UNIVERSITAT DE BARCELONA

Living with Hereditary Cancer Syndromes: Bridging the knowledge gap for inclusive support and comprehensive follow-up

Celia Díez de los Ríos de la Serna

Doctoral Thesis

Living with Hereditary Cancer Syndromes: Bridging the knowledge gap for inclusive support and comprehensive follow-up.

UNIVERSITAT DE BARCELONA

Programa de doctorado en enfermería y salud

Autora: Celia Díez de los Ríos de la Serna

Directoras:

Dra. M.ª Teresa Lluch Canut

Dra. M.ª Paz Fernández Ortega

Tutora: Dra. M.ª Teresa Lluch Canut

Barcelona, 2024

UNIVERSITAT DE BARCELONA

Facultat d'Infermeria

Programa de Doctorado en Enfermería y Salud

Living with Hereditary Cancer syndromes: Bridging the knowledge gap for inclusive support and comprehensive follow-up.

Memoria presentada para optar el grado de doctor por la Universitat de Barcelona

Autora: Celia Díez de los Ríos de la Serna

Directoras:

Dra. M.ª Teresa Lluch Canut

Dra. Paz Fernández Ortega

Tutora: Dra. M.ª Teresa Lluch Canut

Barcelona, 2024

A ti, papá, por llegar a verme doctorada y poder cumplir juntos el sueño.

Les hommes de chez toi, dit le petit prince, cultivent cinq mille roses dans un même jardin… et ils n'y trouvent pas ce qu'ils cherchent… Et cependant ce qu'ils cherchent pourrait être trouvé dans une seule rose ou un peu d'eau…

Le Petit Prince

Antoine de Saint-Exupery

Agradecimientos

Llegar hasta aquí no es fácil. Y sería completamente imposible sin el apoyo y la dedicación de muchas personas que me han ayudado, inspirado y apoyado durante todo este proyecto.

En primer lugar, a las directoras de esta tesis. Que, con sus sonrisas, su apoyo y su disponibilidad me han conducido y ayudado a hacer realidad este proyecto y a hacerlo realidad. A la Dra. Paz Fernández que me animó a seguir persiguiendo el doctorado y que en mis peores momentos me ha apoyado y en los mejores me ha animado y aplaudido. Tenerte de directora, amiga y mentora es un auténtico privilegio. A la Dra. M.ª Teresa Lluch con su atención al detalle, su sonrisa y su sabiduría. Tu entusiasmo y actitud son contagiosos. Hemos vivido y sufrido mucho juntas y me siento enormemente privilegiada de haberos tenido guiándome y apoyándome.

A la Dra. Maura Dowling, que trajo de vuelta la posibilidad de hacer un proyecto europeo. Con su humildad, su seguridad y su paciencia que hacen aprender de ella un auténtico placer.

A todos los amigos y mentores que he conocido y con los que he podido trabajar estos años, de los que he aprendido muchísimo y han hecho de la vida académica un mundo más amable. Especialmente a Greg por enseñarme tanto y darme alas para sentirme segura investigando. Y a mis pollitos por mantenerme cuerda.

A Vicky, Elaine, Alison, quienes creyeron en mi más que yo misma y me animaron a crecer. Por todos los amigos que me han aportado correcciones y sugerencias de mi inglés Vanessa, Beth, Theresa, Mandy y muchos otros.

A Niki, Patrick y Roberta por hacerme entender el poder del Public and Patient Involvement e inspirar mi doctorado para ir un paso más.

A los verdaderos protagonistas de mi historia de vida, sin ellos yo no sería yo. Mi hermano, Juan, mi mayor fan desde mi infancia; mi madre, Belén, la mejor consejera que uno puede tener; y mi padre, Juan, el verdadero mentor de mi vida.

Al mejor compañero de vida que se puede tener, Fábio. Que me ha acompañado, valorado y se ha enorgullecido de cada éxito en nuestra vida personal, laboral y académica. Que ha acogido el doble de responsabilidad para que yo trabajase, viajase, enseñase, sin una sola queja. Que me ha dicho vete a trabajar, estamos bien, estoy orgulloso. Por cada abrazo, cada te quiero y cada ánimo que me ha traído aquí. Por aguantarme, quererme y por enseñarme a diario que todos podemos trabajar en ser mejores personas. Por la paz que me transmites. Por cuidarme.

A mis hijos, que han nacido, crecido y compartido a su madre con este cuarto hijo que es la tesis. Por decirme “pásatelo bien en el trabajo mamá” aun echándome de menos cada día y noche de trabajo. Porque sois maravillosos.

A todos los pacientes que me inspiran a diario, que inspiraron este trabajo y que continúan siendo mi “por qué”.

Y a muchas más personas que me inspiran a diario y me apoyan. Gracias, ¡por tanto!

Contents

Figures

Tables

Abstract

Background: Cancer is one of the leading causes of death worldwide. The most common cancers have a clear relationship with modifiable risk factors such as obesity, alcohol, and tobacco. But the biggest risk factor is having a hereditary cancer syndrome, which significantly increases the risk of cancer relative to the general population. In these individuals, more efforts are needed to decrease and control cancer risk. A personalised approach should consider all known risk factors, including lifestyles, rather than only focusing on early detection techniques. But lifestyles are not sufficiently addressed as a possible cancer risk reduction strategy.

Most behavioural interventions are short, focusing on diet and exercise and producing only short-term benefits that fail to promote broader health literacy or raise awareness of modifiable cancer risk factors. Healthcare professionals do not usually engage in interventions and education to increase health literacy around individual risk and possible actions to reduce it.

Nurses have a unique role in educating patients and their families/caregivers in cancer prevention and risk reduction strategies, which constitute a great opportunity to impact positively on people's health and address cancer-related health literacy. However, nurses miss many valuable opportunities, as they are often unaware of proper risk assessment and risk communication strategies that could benefit patients. This need becomes more acute with hereditary cancer syndrome carriers, as cancer nurses perceive their knowledge around hereditary cancer syndromes as very low. Therefore, they need to acquire additional competencies, preferably within structured educational programmes based on the best evidence and most current professional guidance.

Numerous studies have looked at the attitudes and lifestyles of people affected by hereditary cancer syndromes, but there is little evidence of interventions focused on health promotion. Moreover, research shows that healthcare providers not working in genetics have knowledge gaps regarding cancer and genetics. While there are now ongoing projects to prepare and enhance cancer nurses' knowledge on genetics and genomics, there are no current training programmes available about cancer risk, whether inherited or acquired, or on communication and health promotion.

Aim: The overall aim of this doctoral thesis is to describe the needs of people living with hereditary cancer syndromes and define cancer nurses' educational needs for supporting them.

To this end, a set of **objectives** were defined: (1) to identify which interventions are used to promote healthy lifestyles in people at risk of cancer, (2) to determine what knowledge is necessary for oncology nurses to understand and be able to help people with hereditary cancer syndromes understand their cancer risk and improve their health-related behaviours, and (3) to explore the experience of hereditary cancer syndrome carriers and their priorities and unmet needs during their diagnosis and follow-up.

Methods: The doctoral thesis was planned as a multinational, mixed-methods project, comprising three complementary and consecutive studies to encompass the overall project aim.

The first study, a systematic review of the literature, identified and assessed the effectiveness of interventions used to promote healthy lifestyle behaviours in people with high risk of cancer. The review followed the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines for systematic reviews.

The findings of the systematic review informed a Delphi study, which collected professional perspectives and built consensus about the topics on which cancer nurses need training, specifically in the fields of genetics, hereditary cancer syndromes, and health promotion in oncology. In this study, health professionals from around the world were invited to provide inputs on the activities necessary to promote healthy lifestyles. We used information for the review and published literature and guidelines to create the items for the first round of the Delphi study.

This was followed by a qualitative study, with one-on-one semi-structured interviews with people with hereditary breast and ovarian syndrome or Lynch syndrome from different European countries. Participants underwent individual semi-structured interviews with an interview guide based on relevant nursing and psychology theories around the topics deemed important in the Delphi study. Interviews were recorded, transcribed, and analysed using reflexive thematic analysis.

Results

In the systematic review, four randomised controlled trials were eligible for inclusion. Three included patients with and without a personal history of cancer who were at increased risk of cancer due to inherited cancer syndromes, and one included people undergoing genetic testing due to family history. Interventions targeted three of the known modifiable lifestyle behaviours: diet, physical activity, and alcohol. None included tobacco or any other modifiable lifestyle behaviours.

The international Delphi study involved 74 experts from all around the world including healthcare professionals working in genetics (39%), researchers in cancer and genetics (31%), and healthcare professionals working with cancer patients (30%). A total of 31 items garnered consensus, including knowledge and abilities in genetics, health behaviours, and communication.

The qualitative study included 22 people (8 previvors and 14 survivors) with hereditary breast and ovarian syndrome or Lynch syndrome, from 10 European countries. Analysis of the semi-structured interviews showed similar experiences across diverse countries. Participants' needs were also similar; they reported unmet information and support needs, lack of engagement and actions to inform them on the importance of health behaviours for cancer risk, and a desire to be followed up by cancer professionals. Participants also showed a tendency to access support groups for help and information. The main themes identified were: (unmet) informational and support needs, seeing life in a different way, and limitations of healthcare providers.

Conclusions and recommendations

The evidence generated from the studies indicates a need for cancer nurses to engage in comprehensive follow-up for hereditary cancer syndrome carriers. This follow-up could be crucial to systematically assess individual needs, assess personalised risks, initiate conversations to promote healthy behaviours, and help provide a holistic approach to care that goes beyond surveillance. Whilst surveillance is very important, it is not the only concern for hereditary cancer syndrome carriers. Thus, cancer nurses need to increase their knowledge on genetics and develop knowledge and skills around counselling, communication, and health promotion. The main recommendation emerging from this project is to develop comprehensive follow-up opportunities to support people with HCS, for that it is paramount to develop continuous education programmes for cancer nurses around Europe and to invite them to engage with hereditary cancer syndrome carriers to provide supportive care and follow-up.

Resumen

Introducción: El cáncer es una de las principales causas de muerte en el mundo. Los cánceres más comunes tienen una clara relación con factores de riesgo modificables como la obesidad, el alcohol y el tabaco. El mayor factor de riesgo es tener un síndrome de cáncer hereditario, que aumenta significativamente el riesgo de cáncer en comparación con la población general. En estos individuos, se deben hacer más esfuerzos para disminuir y controlar el riesgo de cáncer. Se recomienda hacer un abordaje individualizado y personalizado utilizando todos los factores de riesgo conocidos, esto debería incluir los hábitos de vida de la persona y no limitarse solo a técnicas de detección precoz. Sin embargo, los estilos de vida no se abordan de forma rutinaria como una posible estrategia de reducción del riesgo de cáncer.

Cuando hay intervenciones centradas en los estilos de vida, la mayoría de las intervenciones se centran en la dieta y el ejercicio, y son de corta duración, lo que da resultados a corto plazo y no promueve la alfabetización sanitaria para concienciar sobre los factores de riesgo de cáncer modificables. Por ello, los profesionales de la salud deben involucrarse más, promoviendo intervenciones y educación con el objetivo de aumentar la alfabetización en salud de sus pacientes sobre sus riesgos individuales y las posibles acciones para reducir su riesgo.

Las enfermeras tienen un papel único en la educación de los pacientes y sus familias/cuidadores sobre las estrategias de prevención y reducción del riesgo de cáncer. Esto ofrece grandes oportunidades para influir en la salud de las personas y promover acciones que reduzcan el riesgo de cáncer. Sin embargo, las enfermeras pierden muchos momentos valiosos, ya que a menudo desconocen cómo valorar y comunicar adecuadamente sobre los riesgos y posibles acciones para reducir estos riesgos. Esta necesidad se hace más evidente con aquellas personas portadoras de un síndrome de cáncer hereditario, ya que las enfermeras tienen un conocimiento limitado sobre los síndromes de cáncer hereditario. Es necesario adquirir competencias adicionales, preferiblemente dentro de programas educativos estructurados basados en la mejor evidencia para orientar a los profesionales y que adquieran estas competencias.

Hay numerosos estudios analizando los estilos de vida de las personas afectadas por síndromes de cáncer hereditario, pero hay poca evidencia de intervenciones centradas en la promoción de la salud y el cambio de estos estilos. También hay evidencia de que profesionales que no trabajan directamente en genética tienen poco conocimiento sobre el rol de la genética en el tratamiento y la prevención del cáncer. Si bien en la actualidad hay proyectos en curso para preparar y mejorar los conocimientos de las enfermeras oncológicas sobre genética y genómica, actualmente no se dispone de educación sobre los riesgos de cáncer en general, tanto hereditarios como adquiridos, para aumentar conocimiento en comunicación y promoción de la salud.

Objetivo: El objetivo general de esta tesis doctoral es describir las necesidades de las personas que viven con síndromes de cáncer hereditarios que aumentan su riesgo de cáncer y definir las necesidades educativas de las enfermeras oncológicas para apoyar a estos pacientes.

Con este fin, se establecieron una serie de **objetivos específicos** : (1) identificar qué intervenciones se utilizan para promover un estilo de vida saludable en personas con riesgo de cáncer, (2) determinar qué conocimientos son necesarios para que las enfermeras de oncología entiendan y puedan ayudar a los pacientes a comprender su riesgo de cáncer y promover un estilo de vida saludable en personas con síndrome de cáncer hereditario, y (3) explorar la

experiencia de los portadores de síndromes de cáncer hereditario y sus prioridades y necesidades insatisfechas durante su diagnóstico y seguimiento.

Métodos: La tesis doctoral se planteó como un proyecto internacional con diferentes metodologías, que comprende tres estudios complementarios y consecutivos para englobar el objetivo general del proyecto.

El primero consistió en una revisión sistemática de la bibliografía para identificar las intervenciones utilizadas para promover comportamientos de estilo de vida saludables en personas con alto riesgo de cáncer y examinar su efectividad. La revisión siguió las guías de Preferred Reporting Items for Systematic Reviews and Meta-Analysis (PRISMA) para revisiones sistemáticas.

A continuación, y derivando de los resultados de la revisión sistemática, se hizo un estudio con metodología Delphi para recoger la perspectiva profesional y llegar a un consenso sobre los temas que los enfermeros oncológicos necesitan en un programa de educación relacionados con genética, síndromes de cáncer hereditario y promoción de la salud en oncología. En este estudio participaron profesionales de la salud de todo el mundo. Para crear los ítems de la primera ronda del Delphi se utilizó información obtenida en la revisión de la literatura y en guías publicadas.

Tras el estudio Delphi, se realizó un estudio cualitativo con entrevistas semiestructuradas a personas con síndrome hereditario de mama y ovario o síndrome de Lynch de diferentes países europeos. Los participantes se sometieron a entrevistas individuales semiestructuradas con una guía de entrevista realizada con base en las teorías relevantes de enfermería y de psicología relacionadas con los temas considerados importantes en el estudio Delphi. Las entrevistas fueron grabadas, transcritas y analizadas mediante análisis temático reflexivo.

Resultados

En la revisión sistemática se incluyeron cuatro ensayos controlados aleatorizados. Tres de ellos incluían pacientes con y sin antecedentes personales de cáncer portadores de síndromes de cáncer hereditario, y uno incluyó a personas que se sometieron a pruebas genéticas debido a antecedentes familiares. Las intervenciones se centraban en tres de los comportamientos de estilo de vida modificables conocidos: la dieta, la actividad física y el alcohol. Ninguno incluyó el tabaco o cualquier otro estilo de vida.

En el estudio internacional con metodología Delphi participaron 74 expertos de todo el mundo, incluyendo profesionales de la salud que trabajan en genética (39%), investigadores en cáncer y genética (31%) y profesionales de la salud con pacientes con cáncer (30%). Un total de treinta y un ítems alcanzaron consenso, incluyendo conocimientos y habilidades en genética, hábitos de salud y comunicación.

En el estudio cualitativo, la muestra incluyó a 22 personas (8 participantes sanos sin cáncer y 14 sobrevivientes) con síndrome hereditario de mama y ovario o síndrome de Lynch de 10 países europeos. El análisis de las entrevistas semiestucturadas mostró experiencias similares independientemente del país de origen de los participantes. Sus necesidades también fueron similares: reconocían necesidades insatisfechas de información y apoyo desde el diagnóstico, una falta de información y de medidas para informarles sobre la importancia de los hábitos de vida saludables para disminuir su riesgo de cáncer y el deseo de que los profesionales de oncología se involucren y realicen un seguimiento regular desde su diagnóstico. Los participantes también mostraron una tendencia a acceder a grupos online de pacientes para buscar apoyo e

información. Los principales temas identificados fueron: necesidades de información y apoyo, ver la vida de otra manera y limitaciones en el conocimiento de los profesionales de la salud.

Conclusiones y recomendaciones Considerando toda la evidencia recogida, es razonable sugerir la necesidad de que las enfermeras oncológicas realicen un seguimiento integral a los portadores de síndromes de cáncer hereditario. Este seguimiento podría ser crucial para evaluar las necesidades individuales, evaluar sus riesgos personales e iniciar conversaciones para promover comportamientos saludables y ayudar a proporcionar un enfoque holístico de la atención que vaya más allá de la vigilancia. Si bien la vigilancia es muy importante, no es la única preocupación para los portadores de síndromes de cáncer hereditario. Para ello, las enfermeras oncológicas necesitan aumentar sus conocimientos sobre genética y desarrollar conocimientos y habilidades en torno a asesoramiento, comunicación y promoción de la salud. La principal recomendación para continuar tras este proyecto es diseñar una formación continua para las enfermeras oncológicas de toda Europa para que se familiaricen con los síndromes de cáncer hereditarios e invitarlas a comprometerse con el seguimiento de las personas con síndromes de cáncer hereditario para proporcionar cuidados de apoyo y seguimiento.

Abbreviations

EONS	European Oncology Nursing Society
ESGH	European Society of Human Genetics
ESMO	European Society of Medical Oncology
HBM	Health Belief Model
HBOC	Hereditary Breast and Ovarian Cancer
HCS	Hereditary Cancer Syndromes
IARC	International Agency for Research on Cancer
ICCN	International Conference on Cancer Nursing
ISONG	International Society of Nurses in Genetics
MMPMH	Multifactorial Model of Positive Mental Health
NCCN	National Comprehensive Cancer Network
NGS	Next Generation Sequencing
PPI	Patient and Public Involvement
PRISMA	Preferred Reporting Items for Systematic Reviews and Meta-Analyses
PROMS	Patient Reported Outcomes Measures
QuADS	Quality assessment with diverse studies
TIDieR	Template for Intervention Description and Replication checklist

Introduction

The International Agency for Research on Cancer (IARC) estimated that there were almost 20 million new cancer cases in 2020, almost 3 million of which were in Europe[1]. Of these new cases, the most common were breast, lung, colorectal, prostate, and stomach cancers[2]. By current estimates, incidence is projected to rise by up to 20% by 2030, making cancer the leading cause of death in the EU[3]. The economic burden of cancer in Europe is a major challenge, as cancer affects the individual economy but also the national health budget and economic growthc[3]. In this context, it is no surprise that every country is investing in efforts to improve cancer care.

In Europe, the Europe's Beating Cancer Plan was conceived to tackle the cancer pathway completely, encompassing prevention, early detection, diagnosis and treatment, and quality of life in cancer patients and survivors[4]. This plan aimed to ensure equal access to all prevention, screening, diagnosis, treatment and supportive care for all EU citizens, but it acknowledged that European countries still face significant disparities, with different cancer plans and differential access to the best available care. According to the European Commission, large inequalities in cancer care persist in different countries and regions, and according to gender, education, income, and age. These inequalities especially affect access to prevention and early detection.

Preventive actions are central, as around 30% to 50% of cancers are considered preventable by avoiding harmful risk factors and adhering to recommendations[5]. Lifestyle behaviours are particularly important, as they are considered a cause of many of the most prevalent cancers and because current evidence suggests that changing behaviours could significantly reduce the cancer burden[6]. Cancer prevention has proven more effective than treatment, constituting the most cost-efficient, long-term cancer control strategy[6]. Prevention strategies can be improved dramatically by raising awareness and addressing risk factors such as tobacco and alcohol consumption, lack of physical activity, obesity, unhealthy diet, extensive sun/sun bed exposure ,and exposure to pollution in the general public and in people already affected by cancer[5,7]. Improving health literacy on cancer risks is the first step to reducing the cancer burden, but worldwide, more efforts have been made in early diagnosis and cancer treatment. A review of the effectiveness of health literacy interventions in Europe showed a variety of interventions with different effectiveness and a big gap compared with other countries like the United States[8].

Over the last decades, the cancer landscape has changed radically with the development of the fields of genetics and genomics. Precision medicine techniques have allowed a better understanding of individual tumours, helping to improve diagnosis and tailor treatments[9]. Treatments have also changed with the adoption of targeted therapies aiming to make cancer more susceptible to treatment and to either cure it or at least

extend survival[10]. Thus, people are living longer with cancer. Precision oncology is also enabling personalised prevention approaches in people with a higher risk of cancer, for example with germline testing of hereditary cancer syndromes, or tertiary prevention in those that have already been affected by cancer, with biomarker monitoring for early prediction of disease progression[11].

Despite the improvements in early detection and advances in treatment, cancer still inspires fear in a large proportion of the population[12]. Much of this fear is rooted in a lack of knowledge around the disease, including ways to avoid it, as almost everyone has either had or known someone who has had cancer.

Moreover, while advances in treatment are helping people to live longer with cancer, mortality is growing in parallel with incidence[1] (Figure 1). In Europe, higher mortality is observed in people with less education and who have cancers related to tobacco and infection, highlighting both the importance of education in prevention and the potential for prevention strategies to reduce the cancer burden[13]. Cancer prevention literacy is therefore a risk factor for worse prognosis.

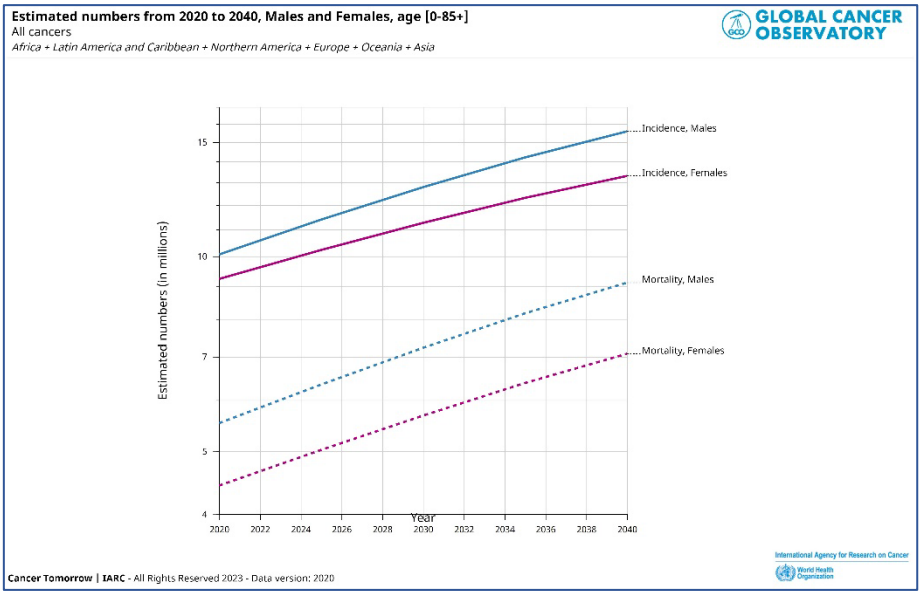

Figure 1. Estimated increase of cancer incidence and mortality. Source: Globocan[2]

Another concerning factor around the latest cancer incidence trends is that while cancer has traditionally been associated with older age, early-onset cancers are on the rise. In people younger than 50 years old, incidence has increased almost 30% in the last two decades and is projected to keep rising[14]. The increase in diagnoses could be influenced by the implementation of screening programmes and early detection techniques[15], but the rising mortality also indicates a true increase in incidence. In younger people especially, these trends are attributed to the lifestyle changes in the general population,

including diet, sedentarism, obesity, pollution, and the microbiome – all of which are believed to interact with genomic and genetic factors[16]. With cases of cancer and mortality increasing overall and in younger populations, cancer control is a priority worldwide[1]. Consequently, healthcare professionals need a good understanding of the possible risks, and they need to be able to engage in conversations with the population around these diseases.

While cancer has been known since the Egyptians, its pathogenesis is still being studied[10]. Defined as an uncontrolled growth of cells, its development is a multi-step process wherein cells undergo genetic changes that affect their behaviour, leading to an excessive proliferation[17]. The gradually improving understanding of how cancer pathogenesis occurs has led to great advances in treatment, but commensurate progress has not been made in prevention. Unlike other common diseases like cardiovascular diseases – still the leading cause of death – cancer is a genetic disease that can affect any part of our body. This makes it more difficult to design campaigns for cancer prevention, and early detection programmes are usually developed according to a single cancer site, for example prostate-specific antigen testing in men or mammograms in women.

Cell changes normally develop over time, and they are influenced by a combination of intrinsic or extrinsic factors such as age, gender, inherited conditions, lifestyle, and environment[18]. Consequently, cancer can affect anyone, but some people are at higher risk than others. The most important factor, dramatically increasing cancer risk, is having an inherited genetic mutation.

Hereditary cancer syndromes

Hereditary cancer syndromes (HCSs), also called inherited cancer syndromes or family cancer syndromes, refer to genetic mutations that can be passed on from parents to children, generally following an autosomal dominant inheritance pattern that alters their risk of cancer[19,20]. The affected genes (normally just one) are usually suppressor genes that work on repairing DNA. These genetic changes can significantly increase the risk of cancer – up to 87% – but the person affected does not always develop cancer[21]. This is because the inherited mutation is only the first step in the pathogenesis of cancer; the cell still has the ability to work normally until external factors, also called somatic mutations, cause the cell to lose its functionality and become a cancer cell[19]. People with HCS carry a higher risk of cancer than those without[19], but once this risk is known, the clinical management can be adapted accordingly.

There are around 50 different HCSs, each involving a number of genes that can be implicated[21–24]. If a HCS is suspected, a single next generation sequencing (NGS) test can be used to look for mutations on multiple genes[22,25]. The panel of genes usually includes a combination of the best known and most prevalent genes for a particular cancer as

well as some suspected but less common genes[26]. This is done to facilitate the identification of genetic alterations that may not have been identified yet, with special relevance for research on genes implicated in hereditary cancer. Although 5% to 10% of the cancers are known to be due to HCSs, the percentage is probably higher, rising up to 20% or even 33% in some studies, if heritability risks are defined as a suggestive family history instead of a specifically identified gene [27,28].

Health professionals can refer to numerous guidelines[29–34] and prediction models [35,36] to decide whether a person should be tested for a suspected HCS. These decision aids take into account information such as age, family history, and cancer disease[24,37]; while they are not infallible, they can help to systematise the testing decisions. Among their limitations, they generally include only the most prevalent genes (such as BRCA 1 and 2 in hereditary breast and ovarian cancer syndromes) and fail to assess other genes commonly available now with the use of NGS panels[24,36]. Their sensitivity may also be limited; for example, the Bethesda guidelines for Lynch syndrome can miss approximately 50% of Lynch carriers[38] and a similar percentage of hereditary breast and ovarian cancer syndrome carriers (HBOC)[39], probably all related to the calculation of only the most prevalent and known genes implicated in HCSs. Still, guidelines and predictive tools serve to facilitate and unify healthcare professional decisions and NGS panels are allowing a better identification of HCS[25] and are especially recommended for healthcare professionals with less experience in genetics.

Together, these rapidly developing tools help professionals and patients to make important decisions to identify carriers, as there are still a large number of unidentified healthy carriers. At the moment, it is estimated that less than 10% of people carrying the BRCA know it[39], and this percentage is much lower for Lynch syndrome despite being even more common[40]. Some authors have pondered whether the population should be routinely screened for HCSs, as identifying these syndromes before a cancer diagnosis offers screening and prevention options[41,42]. The available tools have been developed for the most common HCSs, but not for the many others that exist. Thus, there is an unmet need for comprehensive HCS guidelines in cancer services, and formally trained professionals should be deployed to evaluate the individual's personal and family history. Multidisciplinary boards should hold regular discussion of cases and make testing decisions based on consensus-based criteria, expert discussion, and – when cancer tissue is available – genetic tissue analysis, especially for uncommon HCSs[43].

The most common HCSs are Lynch syndrome, HBOC syndrome, and Li-Fraumeni syndrome[23]. Each HCS increases the risk of cancer at different tumour sites, but they normally have commonalities such as early-onset cancer, several cancers in the same family, or individuals with a personal history of multiple tumours or rare tumours (for example, several cases of ovarian cancer in the same family, or cases of breast cancer in men)[19,20] (Table 1).

Clinical testing is used to find pathogenic variants – those with a known link to an increased risk of cancer. These genetic mutations may be deletions, insertions, large genomic arrangements, or changes in a nucleotide that affect the gene functioning, normally causing premature termination of the gene protein synthesis, which in turn leads to an accumulation of gene alterations and tumour formation[44].

Table 1. Most common hereditary cancer syndromes. Adapted from Garutti et al (2023), Tsaousis et al. (2019), and Gomy and Estevez (2013)[20,22,24]

Syndrome	Genes	Incidence	Anatomical sites with increased cancer risk
Hereditary breast and ovarian cancer syndrome	BRCA1 BRCA2 RAD51 (B,C,D) ATM CHEK2 Only for ovarian BRIP1	1:500 (BRCA1) 1:225 (BRCA2) 1:100 (ATM) 1:937(CHECK2) 1:500 (BRIP1)	Breast cancer Ovarian cancer Pancreatic cancer Prostate cancer Melanoma and others
Lynch syndrome	MLH1 MSH2 MSH6 PMS2 EPCAM	1:279	Colorectal Endometrial Gastric Ovarian Pancreatic Others
Li-Fraumeni syndrome	TP53	1:3500	Brain Breast Ovarian Colorectal Endometrial Melanoma Pancreatic Gastric Prostate Sarcoma Others
Hereditary diffuse gastric cancer syndrome	CDH1	Unknown	Breast Gastric
Peutz-Jegher syndrome	STK11	1:25,000–280,000	Breast Colorectal Pancreatic Polyps
Familial atypical mole-malignant melanoma syndrome	CDKN2A CDK4	Unknown	Melanoma Pancreatic
Cowden's syndrome	PTEN	1:200,000	Breast

Syndrome	Genes	Incidence	Anatomical sites with increased cancer risk
			Endometrial Polyps Skin
Von Hippel–Lindau syndrome	VHL	1:36,000	Pancreatic Renal

Clinical implications of testing

Genetic testing for HCSs seeks to establish an individual's risk of cancer, with the ultimate aim of improving the prognosis of people with HCSs[45]. Once the risk is known, the person is usually enrolled in a cancer screening programme, focused mainly on early diagnosis, risk reduction interventions, and prophylactic surgeries, which encompass curative treatment as well as preventive surgeries to reduce or eliminate the risk[39,46,47]. In addition to risk prevention strategies, HCS testing also represents a gateway to different treatment options[48]. Moreover, it has significant implications for the person's family, namely family cascade testing, posing another challenge for both the HCS carrier and the healthcare system[49].

People with HCSs have reduced life expectancy due to their higher risk of early-onset cancer and cancer in general[50]. Identifying an HCS in a healthy person enables planning for a preventive surveillance programme, including organ-specific surveillance, and prophylactic surgeries that may reduce the risk of cancer, facilitate early detection, and increase the lifespan of the person affected[19,51]. There are surveillance guidelines for the most common HCSs, such as those by the National Comprehensive Cancer Network (NCCN) and the European Society of Medical Oncology (ESMO), which outline testing criteria, risk reduction strategies (including screening and surveillance), and reproductive risks[29–31,34]. These strategies may include an annual colonoscopy for Lynch syndrome or yearly mammograms for HBOC, risk reduction salpingo-oophorectomy for both syndromes, and the use of certain medications, such as tamoxifen in HBOC, that might reduce the risk of cancer. Surveillance becomes more complicated for HCSs that have multiorgan cancer predisposition such as Li-Fraumeni, but even in the most common HCSs, surveillance is focused on the most common cancer risks (like breast and ovarian cancer in HBOC), with less attention to the risk in other tumour sites, such as the pancreas[19,52]. The ESMO guidelines do mention some lifestyle behaviours in the prevention strategies, but NCCN guidelines do not include these recommendations.

While these guidelines give advice on testing criteria, the decision of whether to test or not is usually made by healthcare professionals with knowledge and training in genetics. However, the increased availability of testing has meant that more professionals, especially those working in oncology, are ordering these tests without adequate

knowledge about their implications for cancer management, including family testing and future surveillance and prevention needs[25,53]. These considerations are very important for cancer patients, as the identification of a mutation may entail the possibility of targeted therapies known to be especially effective for these mutations. For example, the incorporation of PARP inhibitors, which are effective against ovarian cancers with BRCA mutations, has increased the life expectancy of these patients[44].

Another important implication of genetic testing is reproductive risk. Identifying an HCS in a person allows for family-wide prevention, as the presence of the same pathogenic variant can be assessed in every family member, but the reproductive risks are related to family members that have not been born yet. Having an HCS entails a 50% chance of passing on mutated genes to offspring[19]. HCS carriers planning on having children need to know the available options in their countries; guidelines include prenatal diagnosis once pregnant or pre-implantation genetic diagnosis using in-vitro techniques[30]. The person also needs to consider the implications of some preventive surgeries, like salpingo-oophorectomy, on their family planning decisions.

HCS testing should include a thorough evaluation and explanation from the healthcare professional, including the interpretation of personal and family history; an exploration of family dynamics; often a physical examination; and explanation of the possible risks, implications of testing, test results, personal and family implications, and risk management strategies[24]. The identification of an HCS should also be accompanied by appropriate counselling. When possible and especially when there is a confirmed HCS or dubious results that have no direct clinical implication for the person affected, people with HCS are referred to genetic counselling. The aim of genetic counselling is to help patients understand and adapt to their individual risk[54,55]. The benefit of genetic counselling in cancer patients is clear, as people affected report better knowledge and reduced anxiety[56,57].

Nowadays, genetic testing is increasing at a faster pace than professionals are being trained as genetic counsellors. Most genetic counsellors are based in the USA, whereas there are scant professionals practicing in other high-income countries and even fewer in low- and middle-income countries[54,58]. Even when genetic counsellors are available, they usually only have two consultations with the person affected – before testing and afterwards, to explain the results, a practice that neglects follow-up needs and may leave patients with unanswered questions about their syndrome[59,60].

The traditional model designed for genetic testing, entailing at least one pre-test and one post-test consultation with a genetic counsellor, is changing[58]. Nowadays, genetic testing in oncology is becoming one more tool necessary for the era of precision medicine, where treatment depends on many factors specific to each patient[61]. NGS has become cheaper and tumour testing more widely available, opening the door to more testing and, more importantly, prioritising the need for results. The traditional model is

moving into the mainstream, where genetic testing is generally done in oncology clinics[62]. Yet, this needed shift has often come at the expense of a comprehensive approach to patient care. Even before the widespread adoption of NGS testing, guidance and follow-up for people with HCS was lacking. Its mainstreaming thus increases the risk of leaving HCS carriers with unmet needs.

Care and management of people with HCSs

All of these clinical, personal, and family implications place important demands on the person tested. One of the most recurring challenges for HCS carriers is the management of uncertainty. When someone is diagnosed with an HCS, they have to cope with an increased risk of cancer, but they do not know when, if, or in which organ they will develop it[63,64]. That probability and feeling of inevitability, without any certainty of what to expect, imposes an emotional burden on HCS carriers[64]. Moreover, being diagnosed with an HCS requires regular follow-ups that may include screening appointments or risk-reducing surgeries, which can make them feel like cancer patients, regardless of whether or not they have had a cancer diagnosis[65,66]. Risk management techniques are not without complications, like early menopause brought on by an oophorectomy, or possible bleeding following colonoscopies with polyps removal[67,68].

Especially in young people, these problems add to worries about their current and possible future offspring. People with children may be plagued with guilt related to the possibility of their children inheriting the HCS from them; those who are thinking of having children also worry about passing on the risk to their children and may struggle to decide how to manage these risks[69,70].

There are other worries for people with HCSs, such as financial toxicity implications. For example, if they want private insurance they need to know if the mutation may alter their coverage, or they may need to plan for the economic burden of the surgery recovery, repeat screening, or planning for the impact that a possible cancer might have[60].

Therefore, genetic testing for HCSs entails the need for proper guidance and help for people affected. While genetic counsellors still have a very important role in oncology, other healthcare professionals need to be able to support and inform patients, as well. Nowadays, HCS carriers are seen by multiple healthcare professionals from primary, secondary, and tertiary care, but they still lack follow-up and a personalised approach. On top of their emotional and physical needs, HCS carriers find healthcare systems difficult. They tend to feel lost and completely alone navigating the system, feeling they have no healthcare professional to turn to in case of doubts[65]. All of these challenges add to the HCS carriers' unmet educational and information needs, affecting their quality of life and self-management decisions and the associated psycho-emotional burden[11,71].

All in all, people with HCSs demand more personalised and centralised care with a reliable healthcare professional to address their needs sensitively and empathically[72,73].

Healthcare professional knowledge and the role of the cancer nurse

In 2019, a European consensus statement and expert recommendations highlighted that the number of genetic counsellors was insufficient for current counselling needs and could not possibly keep up with the rising demand. These experts recommended additional training for oncology professionals as the best strategy to meet the psychological support needs of HCS carriers[62].

Some hospitals are creating services run mainly by cancer nurses in order to address patients' worries and needs[74]. The existing guidelines for the standard of care in HCS carriers state the need to have trained professionals to assist them from diagnosis, but they do not contemplate long-term management needs[43]. By contrast, a model based on a qualitative study on information needs in HCS carriers clearly highlighted follow-up needs[75].

Cancer nurses have a central role and opportunity to fill these gaps[76]. They are essential in promoting person-centred care throughout the disease process[77] and are well positioned to coordinate multidisciplinary care and respond to cancer patients' communication, counselling, and educational needs[78]. However, most nurse-led cancer interventions are related to cancer treatment, while the opportunity to fill the gap in providing preventive services is being missed[78]. Numerous papers have highlighted the importance of involving cancer nurses in the care of HCS carriers[25,26,79], but very few have clearly defined their role and scope of practice. Despite the lack of definition in the healthcare setting, cancer patients and HCS carriers are clear demanding better coordination of care and communication[80].

There is evidence from both inside and outside of Europe that healthcare professionals do not have sufficient knowledge on genetics, and they lack the confidence to adequately support the HCS population[81–84]. Some studies specifically highlight the knowledge needs among cancer nurses[26,85–87]. Without the proper knowledge, healthcare professionals cannot appropriately support HCS carriers' needs, which can lead to emotional distress, over- or under-treatment, and inadequate testing of family members[49].

Cancer nurses could play a crucial role in improving communication and addressing other unmet needs in people with HCSs, especially those related to long-term follow-up and risk-reducing strategies such as surveillance and lifestyle behaviours.

Risk factors: lifestyles

The most prevalent cancers have links to both HCSs and lifestyle behaviours. While HCSs are associated with the largest excess risk, there are also numerous modifiable factors like lifestyle and environment that can influence it[88]. Some are considered modifiable and others partially modifiable, such as as environmental exposures or hormones[7] (Table 2).

Table 2. Cancer risk factors. Adapted from Wu et al. (2018) [18]

Modifiable cancer risk factors	**Partially modifiable cancer risk factors**
Smoking	Radiation
Alcohol	Tumour-causing viruses
Diet	Growth factors
Obesity	Chemical carcinogens
Physical activity	Occupational and environmental factors
Sunlight	Hormones

The World Cancer Research Fund[89] and the International Agency for Research on Cancer (IARC)[90] have undertaken multiple studies and reviews to establish the link between lifestyle behaviours and cancer. Smoking causes almost 20% of cancers[91], obesity contributes to around 8%[92], and alcohol is behind 21.2% of breast cancers and 32.6% of colorectal cancers[93]. There is also an important link between physical activity, sedentarism, and cancer[94]. With the known body of evidence, these global organisations have devised a list of recommendations, including healthy lifestyle behaviours to perform and other behaviours to avoid[95,96] (Figure 2).

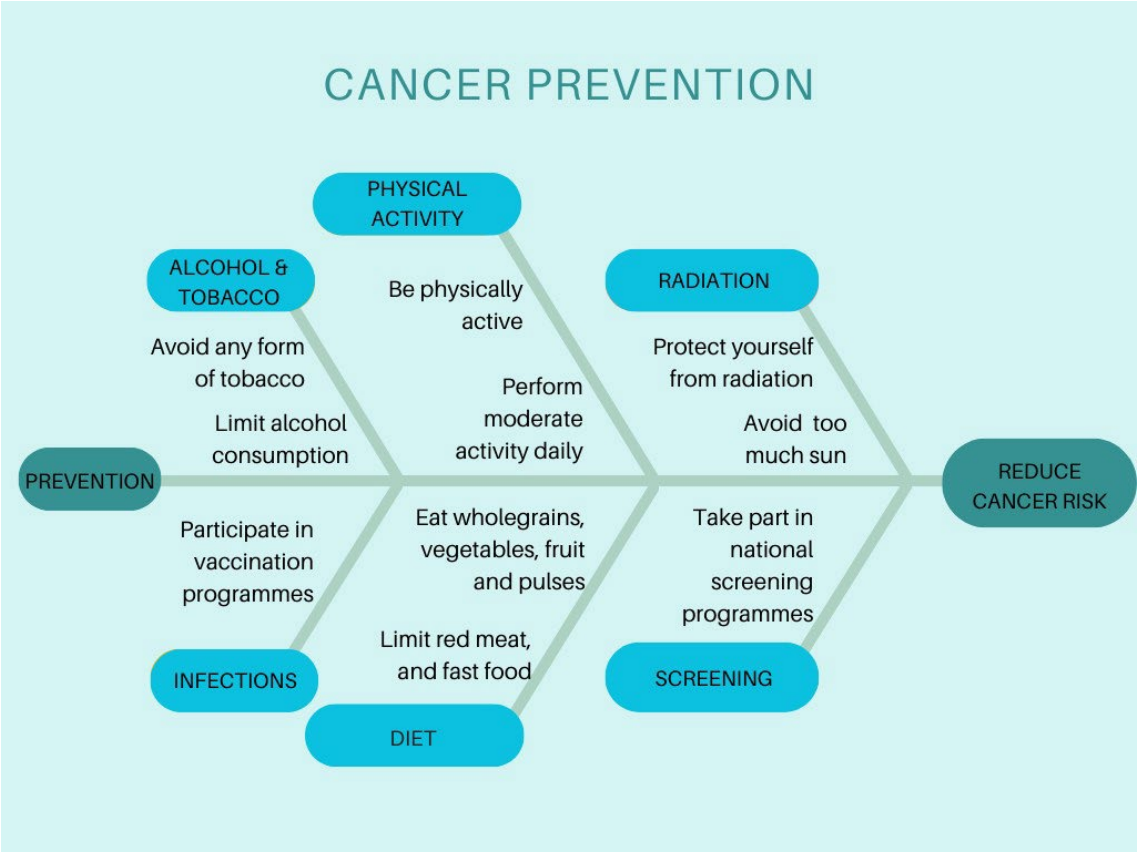

Figure 2. Cancer prevention recommendations. Adapted from the European Code against Cancer and the World Cancer Research Fund[95,96]

If a person already has a genetic susceptibility to cancer due to an HCS, the risk associated with lifestyle behaviours is amplified[5,97]. While there are many possible factors that can cause the genetic changes needed to develop a tumour, behavioural factors could trigger these changes in cells with alterations that make them more susceptible to developing cancer[98]. In Lynch syndrome carriers, obesity increases the risk of colorectal cancer by as much as 49%[99]. At the same time, there are also studies suggesting a risk reduction when adhering to some healthy lifestyle behaviours. For example, aerobic exercise may reduce risk in people with Lynch syndrome by altering the gastrointestinal mucosa[100] and can reduce the risk of breast cancer in BRCA carriers[101,102].

While some HCS prevention and screening guidelines include a section on lifestyle modification, the sections are not very long. And while ESMO guidelines for HBOC do discuss the relevance of regular exercise, weight and alcohol, the recommendations about ovarian cancer are limited to those on oral contraception[30].

HCS carriers usually present two or more lifestyle behaviours considered to be unhealthy[98,103], which they do not necessarily change after HCS diagnosis[104]. In order to

favour the adoption of healthier behaviours, people need to have comprehensive, personalised, understandable information so they can make informed decisions about how to adapt their behaviours to their personal risk[105].

Currently, neither genetic counsellors nor oncology professionals are involved in conversations with HCS carriers about lifestyle. Moreover, while genetic counselling professionals help patients to adapt to their risk and can promote prevention[55], the main focus of genetic counsellors is to help the person to make a decision about genetic testing and then adopt risk-reduction strategies[54], with a focus on cancer screening rather than health promotion[94,97]. Furthermore, there is no clarity on whether lifestyle behaviours are part of their role[55].

Studies assessing the behaviours of people after genetic testing demonstrate that people are more likely to attend cancer surveillance appointments; however, testing alone has little impact on lifestyle behaviours. This situation is likely rooted in patients' scant knowledge on how lifestyle behaviours affect their risk and the little engagement of healthcare professionals in addressing these gaps[106,107].

Addressing lifestyle behaviours

Adequately addressing lifestyle behaviours as a potential risk reduction strategy in people with HCS requires a consideration of health beliefs[107,108]. Knowledge of all the risk factors and beliefs enables a personalised approach that empowers the patient to manage their own risk through an approach known as supported self-management[109,110].

Healthcare interventions can lead to behavioural changes, for example, people attending screening who learn about the effectiveness of healthy lifestyle behaviours for reducing the risk of cancer may adopt beneficial behavioural changes[111]. This has also been observed in cancer survivors[112]. The first need is to raise awareness of the relationship between lifestyles and cancer risk, as many HCS carriers and even healthcare professionals are not well versed on this topic[113–115]. Yet, knowledge alone is insufficient to empower changes; healthcare professionals need to support patients with interventions to promote changes[116,117]. Such interventions can lead to short-term lifestyle modifications in people with HCSs[118], but for these changes to be sustained in time, it is important to also consider and address individual motivations and beliefs[107].

HCS carriers report that healthcare providers often fail to address individual concerns, motivations, and modifiable factors that can affect their risk[71,119,120]. The lack of comprehensive follow-up also affects their care, leaving their needs unmet. Improving this situation requires systemic changes in the healthcare system that enable professionals to adopt a role where the focus is to help people stay healthy by encouraging, facilitating, and supporting self-management. A prerequisite for good care is the competence of professionals in the field who will serve as guide for those affected.

There is a need to increase the knowledge on genetics, risk assessment communication skills, and lifestyle behaviours.

Nurses, and especially cancer nurses, understand the need to keep up with the new advances in oncology, but they have difficulties finding information and courses on genetics[121]. While there are some studies looking at the competencies needed, most are from the USA or Canada[26,85,122]. Likewise, the resources available are from those countries or from the UK, and they address general knowledge in genetics and genomics.

Health education interventions must be based on behavioural theories to promote sustained changes, and none can be effective without good communication skills, a core skill for person-centred care.

Theoretical framework

This project is based on the hypothesis that cancer nurses can address HCS carriers' need for long-term management while supporting and promoting a self-management model of care that encompasses the promotion of healthy behaviours.

At present, there are no healthcare professionals addressing the follow-up of people with HCSs, and moreover there is a lack of understanding on genetics and a lack of involvement in lifestyle behaviours interventions. Thus, it is important to stop and consider the existing theories and models that could inform the planning and implementation of nurses' interventions in this population.

A theory can be defined as "a set of interrelated concepts, definitions, and propositions that present a systematic view of events or situations by specifying relations among variables, in order to explain and predict the events or situations"[123] *(page 26)*. These theories help to shed light on how people learn and change and therefore can guide practice[124].

Numerous conceptual models could potentially set the theoretical groundwork for this thesis. When considering the unmet needs of people with HCS, the theories should help nurses to understand and support patients' worries and concerns; address the person's lifestyle behaviours and promote healthy lifestyles; and empower the person to look after themselves and make informed decisions about their health.

Health Belief Model

The Health Belief Model (HBM) has been widely used in prevention and promotion, as it aims to explain the change and maintenance of health behaviours[123]. It was developed in the 1950s by a group of social psychologists (Hochbaum, Rosenstock and others) in an attempt to explain why preventive behaviours were adopted by some people but not others who received the same information interventions[123].

This model takes into account the effect of an individual's belief and perception on their decisions as to whether or not to modify a health behaviour[125]. It helps to elucidate the different attitudes a person may have when considering a behaviour change, taking into account their perceptions around the likelihood of developing a health problem and its severity, their belief in the effectiveness of specific behaviours for reducing the health problem's effect), and the perceived barriers or negative aspects associated with those behaviours. The HBM also incorporates the perceived self-efficacy (confidence in one's ability to perform the new health behaviour) and cues to action (strategies and factors that prompt action)[123,126].

A worked example of this theory in a person with HCS would consider:

- *Perceived susceptibility*: how large they believe their risk of having cancer is.
- *Perceived severity*: their own beliefs and perception about cancer, for example whether they associate cancer with death, or how serious they see the impact of having cancer on their lives.
- *Perceived benefit*: the person's belief about how beneficial or helpful an action would be, for example a colonoscopy or an exercise routine, for reducing their risk of cancer.
- *Perceived barriers*: the problems that the person perceives around adopting that behaviour such as the preparation of the colonoscopy, travelling to a hospital that they feel far away, or the perceived lack of time to exercising.

Taking this into account is important. A person may strongly believe that they will have cancer, but if they see this as inevitable, if they perceive that the benefit of adopting some healthy behaviour is so small that is not worth it, or if they decide that the barriers of adopting that behaviour outweigh their possible benefit, they may resist preventive interventions. Cues for action capable of changing this calculation could consist of education or a recommendation from the healthcare professional.

While the HBM is not a nursing theory, it can help nursing practice by enabling nurses to devise possible strategies to support and guide patients into preventive actions and healthy lifestyles and to guide intervention planning[127]. The HBM was designed to explain health-related behaviours and has been widely used in public health, including in cancer prevention, for example by increasing uptake of screening for cervical and breast cancer[126]. The theory has also been used to guide the design of preventive interventions and strategies. In research on HCSs, the HBM has been used to understand cancer prevention attitudes, especially in screening, where studies have shown that higher perceived susceptibility, lower barriers to getting screened, higher perceived benefit, and the presence of cues to action affect the attendance to cancer screening[128,129]. Other studies have found that while HCS carriers have high perceived susceptibility, the perceived benefit varies, highlighting the need for healthcare professionals to play a role in education[115,130].

Orem's theory of self-care

Self-care theory is a nursing theory developed by Dorothea Orem in the 1970s, with connections to self-care deficit theory and the nursing system[131]. This theory sustains that there is a necessary connection between the person, nursing, health, and environment, and that nurses can help people to look after themselves, take self-care actions, and recover and confront health problems[131].

- The self-care theory describes how people look after themselves, understanding that self-care consists of behaviour to control factors that affect the person's well-being and prevent possible problems. While it is normally defined in a health situation, there are health deviations of self-care needs. In someone with HCS, this could mean deciding to stay as healthy as possible to either reduce the risk of cancer or being as strong as possible to face it if they get diagnosed.
- The self-care deficit theory explores how nurses can support self-care in a person who either cannot or does not know how to provide it for themselves. Thus, Orem defines what nurses can do to guide, support, or teach the person.
- The nursing system theory describes the relationship between nurses and the person, which can be compensatory, partially compensatory, or supportive, depending on the person's needs.[132]

Orem's theory describes how the nurse can help the person to carry out and maintain self-care actions to maintain their health or to recover or cope with their disease and its consequences. This theory has been widely used in cancer care and has proven effective in increasing cancer patients' ability for self-care[133]. While it has not been as widely applied in cancer prevention, it has been used to increase health literacy, leading to reduced complications and improved quality of life[134]. Health literacy has also been known to be beneficial for cancer prevention[105,129].

If the nursing role can improve health literacy and self-care, this theory would support the nurse's impact on health behaviours and prevention in people with HCSs. This theory has been used to compare the role of nurses in genetics, as in genetic counselling the role of the nurse is to provide enough information for the patient to make an informed decision that could improve the self-care attitudes of the person and their family[135]. Thus, only when HCS carriers have developed behavioural, cognitive, and emotional abilities can they meet the requirements needed for effective self-care.

Lluch's theory on Positive Mental Health

While the concept of positive mental health comes from the 1950s[136], it was Lluch in 1999 who built the Multifactorial Model of Positive Mental Health (MMPMH)[137]. This model defines six interrelated factors that affect a person's ability to cope with health problems:

- *Personal satisfaction*: one's perceived satisfaction in their life and the future.
- *Prosocial attitude*: one's predisposition to be part of the society and support others.
- *Self-control*: emotional balance and the capacity of the person to confront stress and conflict.
- *Autonomy*: the capacity of the person to make their own decisions, together with their self-confidence.
- *Resolution of problems and self-actualization*: one's capacity to analyse, adapt to change and learn, and to develop or grow according to the circumstances.
- *Interpersonal relationship skills*: the ability to connect with others and to have empathy to understand and support others' feelings.

This theory has been used in health promotion and health prevention, as it considers that there is a connection between physical and mental health[137–139] and explores the power that nurses have in promoting self-care skills in this regard. While this theory has not been applied in people with HCS, there have been studies in people with chronic conditions, building a connection between Orem's theory of self-care and the positive mental health theory to show that positive mental health increases the person's self-care abilities[140]. Studies on cancer patients have shown that nurse-led interventions can promote positive mental health, directly influencing the quality of life and patient outcomes[141,142]. This construct and the relationship between self-care and mental health is therefore key for health care in people with HCS, as positive mental health has a direct effect on self-care behaviours, which in turn directly impact health promotion and the person's well-being and quality of life.

Combining the theories for effective nursing interventions

There is a link between a person's capacity for self-care, their health beliefs, and their mental health. Good mental health and a high perceived benefit of personal actions on one's health could have a positive influence on the person's ability to self-care[140]. Moreover, the nurse's support and education could have an impact in all these areas (Figure 3). Nurses have an important role in prevention and health promotion[143,144]. Therefore, they could be the most important healthcare professional to guide and teach HCS carriers to support their self-care abilities.

These theories could explain why HCS carriers do not change their lifestyle and adopt preventive behaviours just because they find out about their increased risk[145]. Rather, the theories support the need for appropriate interventions to promote healthy lifestyle behaviours, based on a good understanding of the individual's needs and beliefs [146].

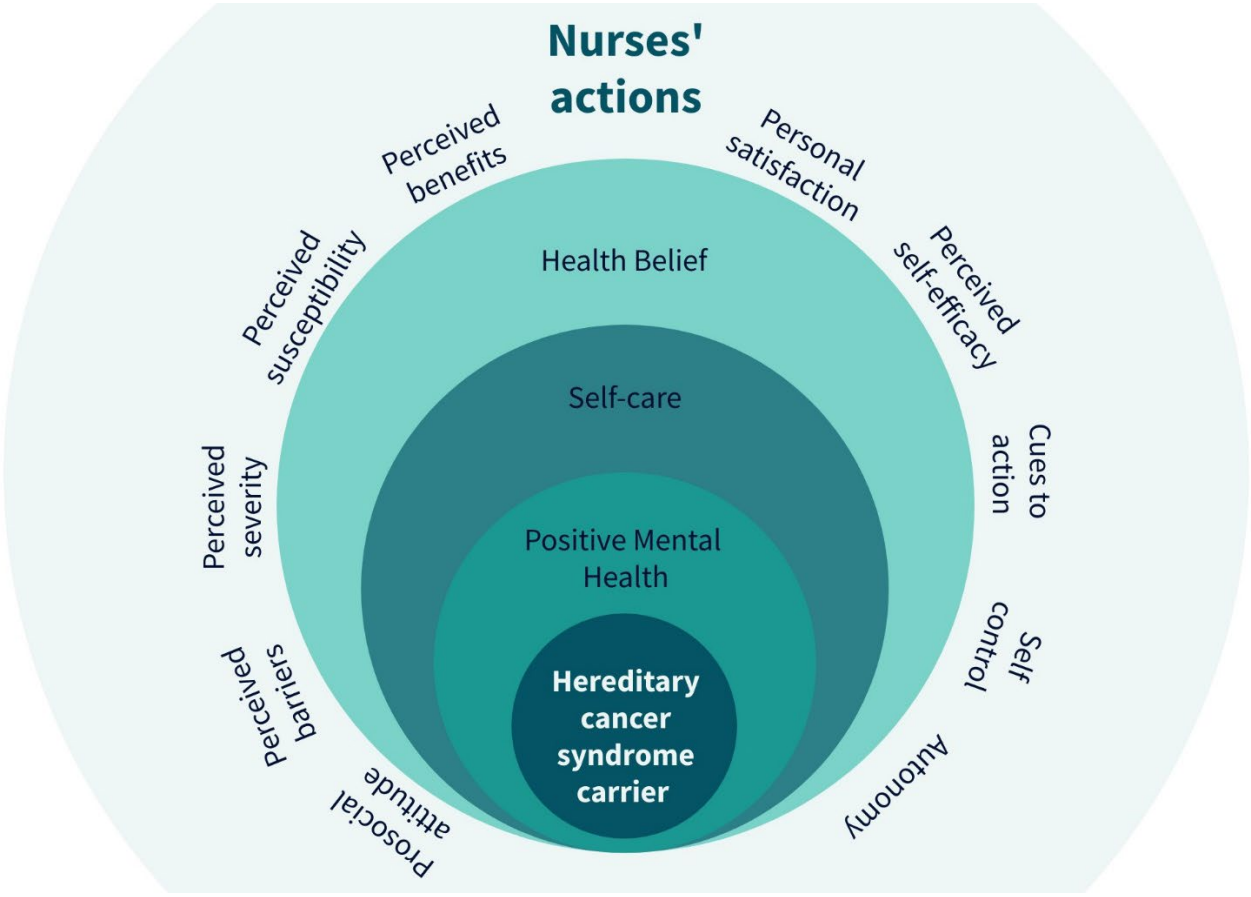

Figure 3. The importance of positive mental health, self-care and health belief in HCS carriers

Patient and public involvement

Patient and public involvement (PPI) is highly recommended in all phases of research, from the identification of the topics to the definition of the relevant questions[147,148]. In this project, patients were actively involved. We recruited two people with HCSs and discussed the proposed research topics in an open forum. HCS carriers provided input on the relevance and usefulness of this topic based on their own experiences or that of their family members. Numerous points arose when discussing the project with them, chief among them the importance of being treated as individuals and being involved so that the research could capture their unique situation. This led to the interviews performed in the project.

Patients were further involved in the project later on. They advised on the design of the project, contributed to the topics covered in the interview guides, and gave their opinion on the data interpretation of the studies. They were also involved in recruiting, allowing the researchers to reach a wider audience.

Aims and objectives

Overarching aim

The overall aim of the study is to describe the needs of people living with hereditary cancer syndromes and define the educational needs of cancer nurses to support them.

Objectives

To this end, a set of objectives was defined:

a) To identify which interventions are used to promote a healthy lifestyle in people at risk of cancer.

b) To determine what knowledge is necessary for oncology nurses to understand and be able to help people with HCS understand their cancer risk and improve their health-related behaviours.

c) To explore the experience of HCS carriers and their priorities and unmet needs during their diagnosis and follow-up.

Objective 1: To identify which interventions are used to promote a healthy lifestyle in people at risk of cancer.

Research Question: How effective are health interventions in promoting healthy lifestyle behaviours according to the literature?

Aims:

- To define which interventions promote healthy lifestyles to reduce risk of cancer.
- To identify the roles and types of intervention activities that have been performed by cancer nurses.
- To identify interventions done in genetic counselling for people with high risk of cancer.

Objective 2: To determine what knowledge is necessary for oncology nurses to understand and be able to help people with HCS understand their cancer risk and improve their health-related behaviours.

Research Question: What are the educational needs that cancer nurses have around cancer and genetics and promoting healthy lifestyle behaviours?

Aim: To reach consensus on desired/required learning outcomes, content, for an educational programme on genetics and health behaviours for cancer nurses.

Objective 3: To explore the experience of HCS carriers and their priorities and unmet needs during their diagnosis and follow-up.

Research Questions:

- What is the experience of HCS carriers from diagnosis through follow-up?
- What unmet needs are they facing and where would they like support?
- What do they think healthcare professionals should know?

Aims:

- To explore the experience of people with HCS.
- To identify the unmet needs while living with HCS.
- To identify the roles, interventions, and educational needs that they demand from healthcare professionals.

Methods

To meet the objectives, a programme of work was planned with a prospective, observational, multinational and mixed-methods design, comprising consecutive studies to answer the different objectives (Figure 4).

The project included a systematic review, a Delphi study, and a qualitative study based on semi-structured interviews.

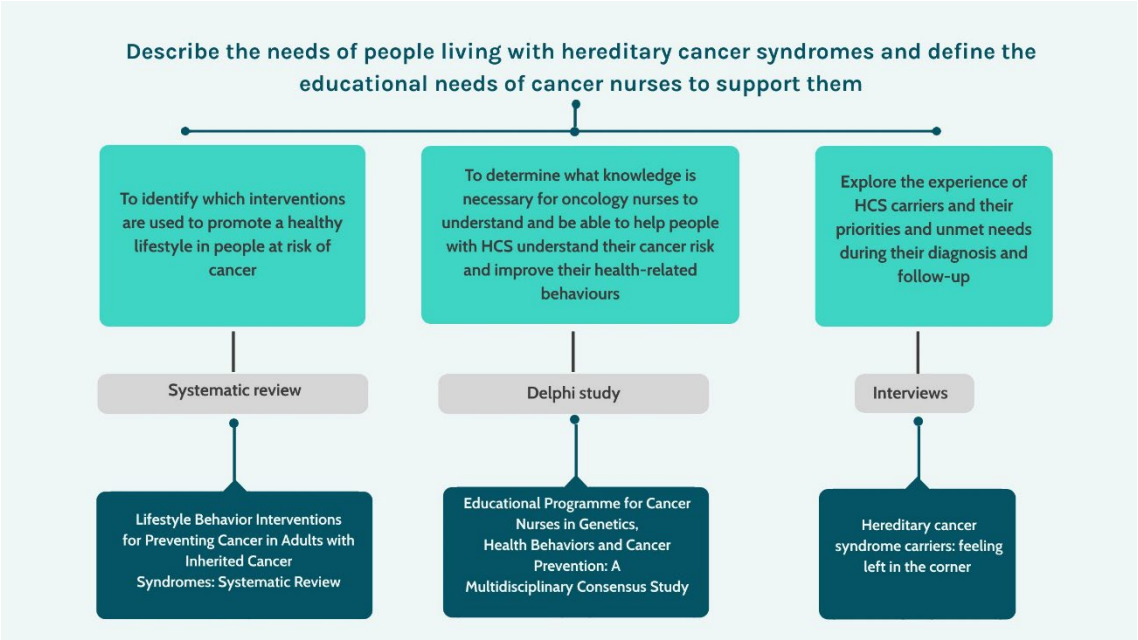

Figure 4. Main aim, objectives, methods and studies derived from the doctoral thesis project

Systematic review

A systematic review was planned to answer objective 1. The review planned to answer the research question and aims derived from objective 1.

A systematic review of the literature was carried out to identify and describe interventions used to promote healthy lifestyle behaviours in people with high risk of cancer. The review included articles from 2010 to July 2022 from the CINAHL, MEDLINE, PubMed, Cochrane Library, Scopus, and Joanna Briggs databases.

The search strategy used terms related to cancer, hereditary cancer and health promotion and included studies focused on adults with high risk of cancer that evaluated effects of behavioural interventions done by healthcare professionals and published in English, Portuguese, or Spanish.

The inclusion criteria included original studies published in peer-reviewed journals, which could be quantitative, qualitative, or mixed methods studies and excluded case studies, expert and medical society recommendations, editorials and commentaries, and studies on patients in active treatment or palliative care.

Analysis: First, one author identified and eliminated obviously irrelevant studies based on the title. For the rest, the abstracts were then obtained and analysed by one author and reviewed by two different reviewers from the research team. For articles that were not identified as irrelevant by all authors, the full texts were obtained and assessed to analyse if they met the inclusion criteria and were relevant.

Data extraction was done in a bespoke Excel sheet and included information like study characteristics, population, lifestyles addressed, description of the intervention, and the measures of effect, based on the Template for Intervention Description and Replication (TIDieR) checklist for reporting interventions[149].

Quality assessment was done using the Joanna Briggs Institute critical appraisal tools for RCTs[150].

The review protocol was published in PROSPERO CRD42020209921 and followed the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines for systematic reviews[151].

Delphi study

The Delphi study planned to answer the research question and aims derived from objective 2.

The Delphi study was performed to collect the professional perspective and reach consensus on the topics on which cancer nurses need training, with relation to genetics, hereditary cancer syndromes, and health promotion in oncology.

Participants: Health professionals from around the world were invited to participate to gather the professional perspective of the activities necessary to promote healthy lifestyle habits. The inclusion criteria were: healthcare professionals with experience in the care/treatment of people with cancer or people at high risk for cancer, and able to understand English or Spanish.

Sample size: The Delphi methodology requires approaching experts as panel members. Convenience sampling was used. This study planned to recruit a minimum of 36 participants. While there is no defined number for a Delphi study, the panel needs to be large enough to represent consensus, and there are recommendations to recruit around 18 experts to ensure sufficient contributions; assuming an attrition rate of 50%, a sample of 36 participants was sought [152,153].

Recruitment was done with direct invitations to expert authors of papers relevant to the

topic and to members of the European Society of Human Genetics, Cancer Genetics Group, the International Society of Nurses in Genetics, and the Global Genomics Nursing Alliance group. Experts were also invited through social media and expert groups, and snowball sampling was used, as we asked experts to share the invitation with other experts in the field.

The Delphi study was carried out using the university Microsoft Forms in order to comply with data protection regulations and guarantee the privacy of the volunteers participating in the research and their personal data, keeping the information secure on the university servers.

The Delphi study initially included 18 potential topics identified from the systematic review, the theoretical frameworks that sustain this project, and the EONS Cancer Nursing Education Framework[154]. Items were divided in 3 domains: genetics, lifestyle behaviours, and communication and barriers. In round 1, experts assessed the relevance of topics identified during the systematic review and suggested additional terms. The following rounds aimed to reach consensus on the important topics by including or excluding topics that reached 75% of consensus.

Round 1 consisted of three sections:

Section 1: Baseline demographic characteristics (e.g., age, gender, country, function, and clinical role). Participants' email addresses were requested for maintaining contact during the different rounds.

Section 2: Participants were asked to rate the importance of topics for inclusion in the programme on a 7-point Likert scale (1–3 = not important, 4= unsure, 5–7 = important).

Section 3: Open-ended comment boxes for participants to suggest additional topics that were not mentioned in the original list.

The second-round items were sent to all first-round responders to reassess the importance of the points that achieved consensus agreement, consensus neutrality, and discord in the previous round. All the final results and final comments were shared with the participants and opened for comment.

Data analysis: The results of the first round were exported to an Excel sheet and anonymised for analysis by the three members of the research team. Analysis of the Likert scale was done by descriptive analysis. The percentage of agreement of important, unsure, and not important was measured. Agreement of 75% of participants was defined as consensus[155]. Content analysis was used by the authors to analyse comments and suggest additional items. Items were grouped into themes and were either incorporated into existing competencies or formulated as new competencies.

Semi-structured interviews

A qualitative study was planned to answer objective 3. The semi-structured interviews planned to answer the research question and aims derived from objective 3.

Method: Qualitative study with semi-structured interviews designed to cover the areas of interest but ensuring that the flow of the interview was directed by the participant.

Participants: Hereditary cancer syndrome carriers with either hereditary breast or ovarian syndromes or Lynch syndromes, regardless of a previous diagnosis of cancer from around Europe, and able to speak in Spanish or English. People who were undergoing cancer treatment at the time of the study, or who were not confirmed carriers of an HCS, were excluded.

Sample size: Theoretical sample selection was calculated to allow diversity of participants regarding their age, country of origin and cancer syndrome. Within qualitative research, the goal is to select participants who can provide extensive information about the phenomena being studied and not to generalise the findings of a randomly selected sample. For this study, a theoretical sample size of 15-20 individuals, including people with both Lynch syndrome and HBOC, from a demographically and geographically heterogeneous European sample was planned.

Recruitment: Recruitment was done through posts on social media inviting eligible participants to contact the principal investigator. The study allowed for snowball sampling, where some participants knew and shared the study with other HCS carriers.

As the participants were recruited from all over Europe, participants were invited to a semi-structured online interview lasting around 40 minutes. The interviews were done in English or Spanish following participants' preferences and used an interview guide to explore their experiences during the diagnosis of their HCS; their experiences with professionals during follow-up; how participants perceived and managed their risk in terms of lifestyle, mental health, and communication with healthcare professionals; the recommendations obtained; their unmet needs and sources of information; what they felt helped them and what didn't; and what difficulties could have been made easier had health professionals had more knowledge. The interview guide was created based on the Health Belief Model[123], Orem's theory of self-care[156], and Lluch's theory on positive mental health[137].

Interviews were recorded, and anonymised transcripts were used for analysis. Field notes, comments, and observations were added to the transcription when the PI reviewed them.

Data analysis was done using reflexive thematic content analysis on the verbatim transcript, following the six-phase process: reading and familiarization, coding, searching for themes, reviewing themes, defining and naming themes, and finalizing the

analysis[157,158]. For the analysis, a coding frame was created by the researchers[159] and then further analysed. Some participants reviewed the themes and quotes and gave feedback to refine the analysis.

Ethical approval

As this study recruited participants from around Europe but without using other centres as recruiting centres, the doctoral student requested and obtained ethical approval from the Ethical Committee for Clinical Research of University of Barcelona (IRB00003099; Annex 1).

Every participant that expressed interest to be part of the Delphi had access to the participant information and was asked to complete the consent form online prior to answering the first Delphi round.

Participants that expressed interest in being part of the semi-structured interviews were sent the participant information and consent form via email. They had to return the consent form prior to scheduling the interview dates. Even if they signed a consent form, participants were reminded of the objectives of the research and were asked for oral consent to start recording the interviews.

The participant information and consent forms are included in Annex 2.

Results

Article 1:

Title	Lifestyle Behaviour Interventions for Preventing Cancer in Adults with Inherited Cancer Syndromes: Systematic Review.
Authors	Diez de los Rios de la Serna, C., Fernández-Ortega, P., Lluch-Canut, T
Reference	**Diez de los Rios de la Serna C**, Fernández-Ortega P, Lluch-Canut T. Lifestyle Behavior Interventions for Preventing Cancer in Adults with Inherited Cancer Syndromes: Systematic Review. *Int J Environ Res Public Health*. 2022;19(21).
Journal	International Journal of Environmental Research and Public Health.; 19(21):14098.
Keywords	Cancer prevention; lifestyle intervention; hereditary cancer; risk reduction; high-risk cancer; health behaviors
Impact Factor	3.39
Category	Public Health, Environmental and Occupational health
Quartile	Q2
ISSN	1660-4601

International Journal of
Environmental Research and Public Health

MDPI

Review

Lifestyle Behavior Interventions for Preventing Cancer in Adults with Inherited Cancer Syndromes: Systematic Review

Celia Diez de los Rios de la Serna [1,*,†], Paz Fernández-Ortega [1,2] and Teresa Lluch-Canut [1]

[1] School of Nursing, Faculty of Medicine and Health Sciences, Bellvitge Campus, University of Barcelona (UB), 08907 Barcelona, Spain
[2] Institut Català d'Oncologia (ICO) Barcelona, Bellvitge, 08908 Barcelona, Spain
* Correspondence: cdiezdde7@alumnes.ub.edu or celiadiezrios@gmail.com
† PhD candidate.

Abstract: (1) Background: The link between lifestyle behaviors and cancer risk is well established, which is important for people with personal/family history or genetic susceptibility. Genetic testing is not sufficient motivation to prompt healthier lifestyle behaviors. This systematic review aims to describe and assess interventions for promoting healthy behaviors in people at high risk of cancer. (2) Methods: The review was performed according to PRISMA guidelines using search terms related to hereditary cancer and health education to identify studies indexed in: CINAHL, MEDLINE, PubMed, Cochrane Library, Scopus, and Joanna Briggs, and published from January 2010 to July 2022. (3) Results: The search yielded 1558 initial records; four randomized controlled trials were eligible. Three included patients with and without a personal history of cancer who were at increased risk of cancer due to inherited cancer syndromes, and one included people undergoing genetic testing due to family history. Interventions targeted diet, physical activity, and alcohol. (4) Conclusions: There is a paucity of research on interventions for promoting healthy lifestyle behaviors in people with a high risk of cancer. Interventions produced positive short-term results, but there was no evidence that behavioral modifications were sustained over time. All healthcare professionals can actively promote healthy behaviors that may prevent cancer.

Keywords: cancer prevention; lifestyle intervention; hereditary cancer; risk reduction; high-risk cancer; health behaviors

Citation: Diez de los Rios de la Serna, C.; Fernández-Ortega, P.; Lluch-Canut, T. Lifestyle Behavior Interventions for Preventing Cancer in Adults with Inherited Cancer Syndromes: Systematic Review. *Int. J. Environ. Res. Public Health* **2022**, *19*, 14098. https://doi.org/10.3390/ijerph192114098

Academic Editor: María M. Morales Suárez-Varela

Received: 10 September 2022
Accepted: 25 October 2022
Published: 28 October 2022

1. Introduction

Cancer is a multifactorial disease resulting from a combination of genetic and external factors [1], and it is projected to eventually become the leading cause of death in every country in the world [2].

Cancer has a clear relationship with modifiable risk factors such as obesity, alcohol, and tobacco [3], and with partially modifiable factors such as environmental exposures and hormones [4]. Around 5–10% of the population has a very high risk of cancer due to inherited mutations [5], and in this group, the relationship between modifiable risk factors and cancer is more pronounced than in the general population [6–8] (Figure 1). For example, obesity can increase the risk of colorectal cancer by 49% in people with a genetic mutation [7]. Likewise, a systematic review found that drinking alcohol and being overweight increased breast cancer risk in BRCA carriers, while physical activity reduced it [9]. A prospective cohort study estimated that physical activity can reduce breast cancer risk in women with BRCA1 and BRCA2 by approximately 20% [8]. Therefore, modifying non-genetic risk factors related to behaviors or hormones can help decrease the relative risk of cancer [10].

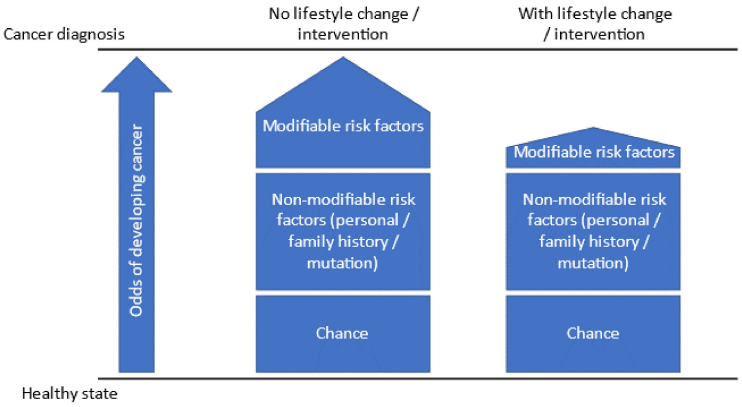

Figure 1. Addressing behavioral factors in people with increased risk for cancer can decrease their odds of developing cancer.

Cancer prevention interventions constitute the best approach for reducing incident cases and known risk factors, and in turn the morbidity and mortality of some diagnoses [11,12] (Figure 2). However, these campaigns tend to be population-based rather than targeted to risk groups. Some patients' associations and organizations for people affected by inherited cancer syndromes such as Facing Hereditary Cancer Empowered (www.facingourrisk.org, accessed on 9 September 2022) or AFALynch (afalynch.org, accessed on 9 September 2022) do organize campaigns and programs to improve health literacy, with the main aim of enabling people to make healthier lifestyle decisions and empower them to manage their personal cancer risk.

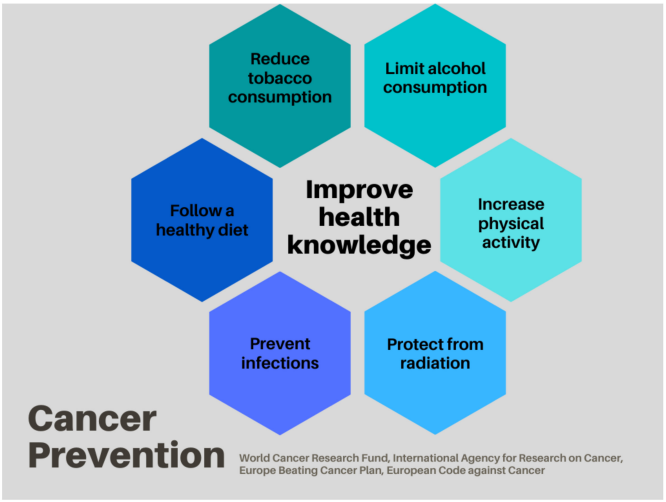

Figure 2. All cancer prevention strategies described can be grouped into these strategies. The figure is based on recommendations from the World Cancer Research Fund, International Agency for Research on Cancer, Europe Beating Cancer Plan, and the European Code Against Cancer [13–16].

1.1. Genetic Counseling

Those who carry an increased risk of cancer due to their personal or family history are normally referred for genetic counseling for predictive testing. According to the Transnational Alliance of Genetic Counseling, the main aim of these consultations is to help patients understand their individual risk and make a decision about whether genetic testing is appropriate for them [17]. Counselors may also assess patients' lifestyle and educate them on how to adapt to their cancer risk by reducing behavioral components [18], although there is no consensus on their precise role in providing advice about lifestyle behaviors [18].

Indeed, when a patient has an inherited cancer syndrome, genetic counseling focuses more on cancer screening and preventive surgery than on health education [19–21]. Thus,

following these consultations, patients are more likely to increase their cancer surveillance or opt for risk-reducing surgeries than to change behaviors [22]. The studies do not explain whether this is due to lack of awareness and information provided during counseling or because these interventions reduce their perception of risk [21]. A systematic review [22] of lifestyle behaviors in patients receiving genetic counselling found that communicating the risk of cancer due to genetic alteration has little impact on lifestyle behaviors such as smoking, diet, or physical activity. However, a review evaluating interventions during colorectal and breast cancer screening found that behavioral interventions can promote increased physical activity and dietary modifications [23]. The same tendency has been observed in cancer survivors, who are motivated to engage in interventions following treatment; however, these changes are not normally sustained long term [24,25].

1.2. Changing Lifestyle Behaviors

Using all the information of an individual's known risk factors, including their behavioral habits, is necessary for a personalized approach. Assessing individuals' risks, motivations, and priorities gives people the opportunity to self-manage their risk [6]. However, knowledge alone is insufficient for effective cancer prevention; it must be supplemented with health education interventions that favor behavior change [26].

People with an increased risk of cancer seek advice from different healthcare providers, but these professionals may miss opportunities to provide information and motivate individuals to change health-related behavior [27]. Family physicians, nurses, and other health professionals often lack proper risk assessment and communication skills [21]. The precise impact of health interventions on health behavior in patients at high risk of cancer due to inherited cancer syndromes remains unquantified.

Improving awareness on this important topic would support the identification and planning of interventions tailored to these individuals' needs and empower them to reduce risky behaviors, thereby improving overall cancer morbidity, survival, and the patient experience.

This systematic review was conceived to address this gap in knowledge by identifying and evaluating interventions for promoting healthy lifestyles in people with a high risk of cancer due to inherited cancer syndromes.

The primary aim is to assess the effect of health education interventions for modifying lifestyle behaviors in adults with a high risk of cancer. The secondary aim is to identify the healthcare professionals responsible for the interventions and describe motivations and barriers for change.

2. Materials and Methods

This systematic review followed the Joanna Briggs Institute methodological guidelines [28] and was reported according to the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) statement guidelines [29]. The review was registered in 2020 on PROSPERO: CRD42020209921 (PRISMA checklist included in Supplementary Table S1).

The research question was formulated using the PICO typology [30]: P—Population: adults at increased risk of cancer; I—Intervention: health education interventions; C—Comparison: no intervention; O—Outcome: modification of lifestyle behaviors.

Patient and public involvement: Input from public and patient involvement informed this research. Specifically, three people with genetic syndromes (one with BRCA unaffected by cancer, and two with Lynch syndrome—one with cancer and the other without) were involved in the project development phase and provided feedback on the appropriateness and pertinence of the objectives to the population under study.

2.1. Search Strategy

A systematic search was conducted in the following electronic databases: Ebsco CINAHL, Ovid MEDLINE, PubMed, Cochrane Library, Scopus, and Joanna Briggs. Relevant peer-reviewed studies published from January 2010 to August 2022 were included,

as genetic counseling related to cancer risk only began in the late 1990s, and published studies on behavioral risks in these populations did not begin to appear until the 2010s [18]. References cited in systematic reviews evaluating lifestyle interventions [23,31,32] in other populations were screened for additional articles which might have been overlooked. The website clinicaltrials.gov was also checked for any published protocols or feasibility studies.

The search strategy combined the key PICO terms using free text and MeSH terms related to cancer, hereditary cancer, and health education and promotion (Supplementary Table S2). A university librarian was consulted to validate the search strategies.

The searches were limited to research articles published in English, Portuguese, or Spanish (see Supplementary Table S3 for an example of a database search).

2.2. Eligibility Criteria

All intervention studies that met the inclusion criteria and were published in peer-reviewed journals were evaluated.

Inclusion criteria were based on the research question and study objectives:

- Studies focused on adults with a high risk of cancer, defined as those with a significant personal or/and family history of cancer undergoing genetic testing or confirmed inherited cancer syndrome [5].
- Studies evaluating the effects of behavioral interventions.
- RCTs and other experimental studies researching the effect of health education interventions in this population (randomized trials and non-randomized trials) with or without a control group (experimental studies comparing the intervention vs another form of intervention as comparator), and written in English, Portuguese, or Spanish.
- Articles were excluded if they were:
- Studies of unmodifiable factors such as genes.
- Studies not assessing behavioral interventions (for example evaluating the effect of medication or screening).
- Studies in people receiving active treatment for cancer, as they experience different cancer-related barriers and have different motivation towards interventions that improve quality of life or symptoms rather than reducing risk [33].
- Expert and medical society recommendations, editorials, reviews, and commentaries.
- Study protocols, case reports, or drug trials.
- Studies performed only in animals.
- Studies that exclude patients with genetic mutations.

2.3. Screening

The principal investigator (PI) performed an initial screening of titles for all records retrieved by the search. Potentially relevant publications were downloaded into reference management software and de-duplicated. The PI screened the abstracts against the eligibility criteria, and then two authors independently read the full text of the remaining articles to determine whether they met the review's inclusion criteria.

2.4. Assessment of Methodological Quality and Bias

The Cochrane RoB2 tool was used to assess the risk of bias in the included RCTs [34]. This tool is used to rate each specified outcome as being at low risk, causing some concerns, or having a high risk of bias. Quality was assessed using the Joanna Briggs Institute critical appraisal tools for RCTs [35]. No studies were excluded based on these assessments.

2.5. Data Extraction and Synthesis

The PI extracted data into a customized evidence table in Excel, and the second author double-checked them. The data extraction form was piloted using the first studies to define what information to collect and ensure comprehensive data capture. Data included study characteristics, population, lifestyles addressed, description of the intervention, and the measures of efficacy, based on the Template for Intervention Description and

Replication (TIDieR) checklist for reporting interventions [36]. In addition, we noted which professionals delivered the intervention along with motivations and barriers for behavioral modification and engagement.

The results were combined in an organized, visual table, where comparable results can be pooled as recommended by the Centre for Reviews and Dissemination (CRD) guidance for undertaking reviews in healthcare [30].

Intervention studies involving animals or humans, and other studies that require ethical approval, must list the authority that provided approval and the corresponding ethical approval code.

3. Results

The initial database search yielded 1558 records. After screening titles and abstracts, 51 publications were retrieved for full-text review, and four RCTs met our selection criteria (PRISMA flow charts; Figure 3).

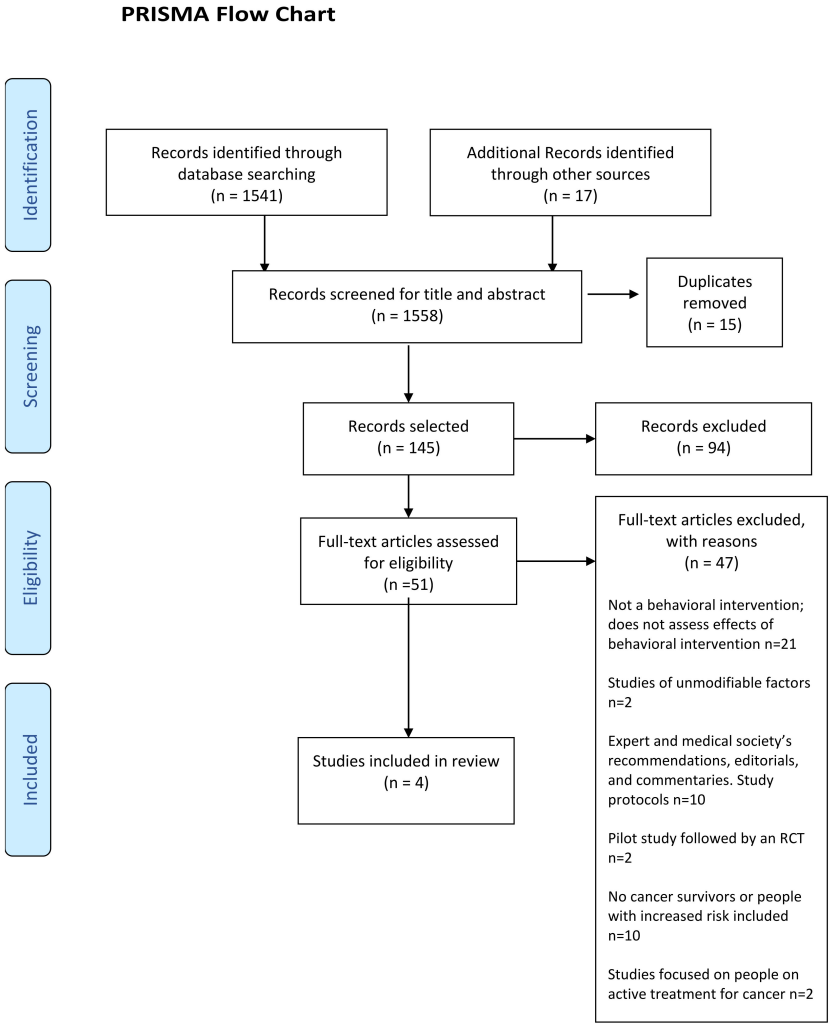

Figure 3. PRISMA diagram of the article selection/screening process (adapted from [28]).

3.1. Characteristics of Included Studies

Table 1 summarizes the main characteristics of included studies, which all took place in Europe (one each in the UK, Germany, The Netherlands, and Italy). All were written in English.

Table 1. Characteristics and methodology of the studies.

Study	Design, Country	Sample Size	Population	Intervention	Comparator	Duration	Outcomes Primary	Secondary	Measurements
Anderson et al., 2018 [38]	2-arm RCT (feasibility), UK	N = 78 (intervention n = 39, control n = 39)	People with family history of breast or colorectal cancer and BMI of $\geq$25 kg/m^2	Face-to-face session plus 4 telephone consultations, pedometer, and walking program	Usual care	3 months	Feasibility measures	Changes in weight, physical activity, diet, psychosocial measures	Changes in weight: kg, waist circumference and BMI. Physical activity: IPAQ-Short and physical activity monitors (with sedentary time, moderate and vigorous activity, and step counts) Diet: Dietary Instrument for Nutrition Education questionnaire Alcohol: 7-day alcohol record
Kiechle et al., 2017 [37]	2-arm RCT(feasibility), Germany	N = 68 (intervention n = 33, control n = 35)	*BRCA1* or *2* carriers with cancer	Structured face-to-face behavioral intervention for increased physical activity and nutrition education	Lecture on the positive effects of PA and healthy diet	12 months (3 intervention, 9 supervision)	Adherence to and acceptability of the intervention	Effects on physical activity, diet, BMI, QoL, and stress	BMI Diet: MEDAS Questionnaire and eating habits, nutrient and fat calorie intake (EPIC-FFQ) Physical activity: maximal oxygen intake (VO_2 peak), ventilatory threshold (O_2 at VT1), and physical activity (IPAQ)

Table 1. *Cont.*

Study	Design, Country	Sample Size	Population	Intervention	Comparator	Duration	Outcomes Primary	Secondary	Measurements
Vrieling et al., 2018 [40]	2-arm RCT, Netherlands	N = 226 (intervention n = 114, control n = 112)	People with Lynch syndrome with and without cancer	WCRF health promotion materials and information about colorectal cancer symptoms and prevention	Usual care	6 months (1 intervention, 5 follow-up)	Awareness of cancer risk factors	Adherence to WCRF recommendations	BMIWCRF/AICR adherence, Diet: adapted version of FFQ validated questionnaire Physical activity: Short Questionnaire to Assess Health Enhancing Physical Activity (SQUASH)
Bruno et al., 2020 [39]	2-arm RCT, Italy	N = 502 (intervention n = 254, control n = 248)	BRCA carriers, with or without a previous cancer	Dietary activities, cooking courses followed by lunch and nutritional conferences	Recommendations on cancer prevention	6-month intervention	IGF-I reduction	Food intake	Height and body weight Diet: MEDAS Questionnaire

AICR: American Institute for Cancer Research, BMI: body mass index, FFQ: food frequency questionnaire, IGF-I: insulin-like growth factor-I, IPAQ: International Physical Activity Questionnaire, MEDAS: Mediterranean Diet Adherence Score, RCT: randomized controlled trial, QoL: quality of life, WCRF: World Cancer Research Fund, VT1: ventilatory threshold 1.

The samples sizes ranged from 29 to 502 adult participants, with a mean age of 41 [37] to 49 years old [38] (range 24 to 72). One study's (25%) primary outcome was changes in lifestyle behavior. Kiechle et al. [37] and Bruno et al. [39] studied patients with the BRCA mutation, Vrieling et al. [40] studied patients with Lynch syndrome, and Anderson et al. [38] included people with a family history of breast or colorectal cancer prior to genetic testing. One study included only healthy individuals with a high risk of BRCA or Lynch [38], one only BRCA carriers with a personal history of cancer [37], and the other two populations with hereditary alterations (Lynch syndrome [40] and BRCA carriers [39]), with or without a personal history of cancer.

3.2. Overall Methodological Quality of the Studies and Risk of Bias of the RCTs

The methodological quality of the studies was particularly affected by the lack of blinding in participants and in the professionals delivering the intervention. It was also unclear whether the assessors were blinded to the trial arm (Table 2).

Table 2. Quality assessment using the Joanna Briggs Institute critical appraisal tools for RCTs [35].

Questions for Critically Appraising the Quality of RCTs	Anderson et al. [38]	Kiechle et al. [37]	Vrieling et al. [40]	Bruno et al. [39]
1. Was true randomization used for assignment of participants to treatment groups?	Unclear	Yes	Unclear	Unclear
2. Was allocation to treatment groups concealed?	Yes	Yes	Unclear	Unclear
3. Were treatment groups similar at baseline?	Yes	Yes	Unclear	Yes
4. Were participants blind to treatment assignment?	No	No	No	No
5. Were those delivering treatment blind to treatment assignment?	No	No	No	No
6. Were outcomes assessors blind to treatment assignment?	Yes	Unclear	Unclear	Unclear
7. Were treatment groups treated identically other than the intervention of interest?	Yes	No	Yes	Yes
8. Was follow-up complete and if not, were differences between groups in terms of their follow-up adequately described and analyzed?	Yes	Yes	Yes	Yes
9. Were participants analyzed in the groups to which they were randomized?	Yes	Yes	Yes	Yes

Table 2. *Cont.*

Questions for Critically Appraising the Quality of RCTs	Anderson et al. [38]	Kiechle et al. [37]	Vrieling et al. [40]	Bruno et al. [39]
10. Were outcomes measured in the same way for treatment groups?	Yes	No	Yes	Yes
11. Were outcomes measured in a reliable way?	Yes	Yes	Yes	Yes
12. Was appropriate statistical analysis used?	Unclear	Yes	Unclear	Yes
13. Was the trial design appropriate, and any deviations from the standard RCT design (individual randomization, parallel groups) accounted for in the conduct and analysis of the trial?	Unclear	Yes	No	Unclear

The risk of bias assessment showed that all RCTs either caused some concerns or were at high risk of bias (Figure 4).

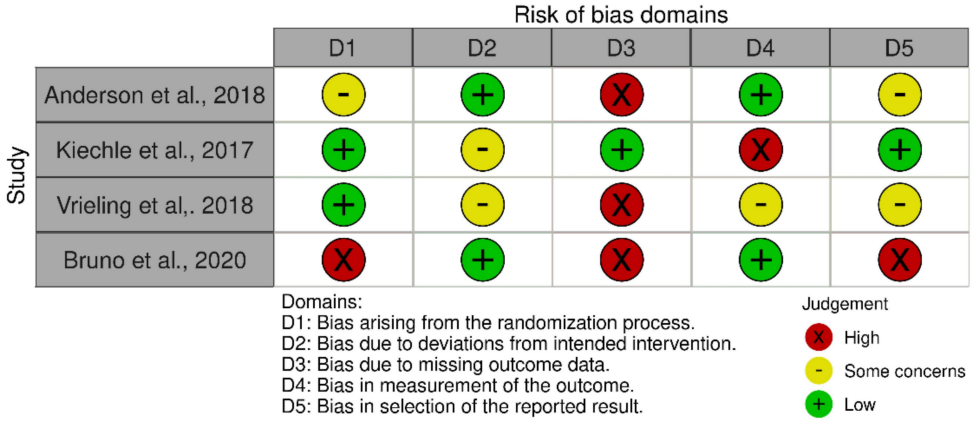

Figure 4. Risk of bias of included studies [14] Studies: Anderson et al. [38], Kiechle et al. [37], Vrieling et al. [40] and Bruno et al. [39].

Randomization and allocation concealment were reasonably well described for most studies, as were outcomes and reasons for participant attrition. Logically, most participants were aware of the intervention they were allocated to, but most studies did not clarify if the outcomes assessors were blinded to treatment assignment.

3.3. Interventions

All four included studies that targeted diet, while three also assessed interventions to increase physical activity. Alcohol intake was targeted in two studies but was not reported in either. One study mentioned tobacco but did not report results.

Regarding the type of intervention, one study used a combination of dietary activities and cooking classes [39]; one, information delivered by leaflets [40]; and two, information provided through consultations and education [37,38].

Interventions were somewhat different in terms of the mode of delivery, duration, and the nature of the intervention. All four studies had some kind of face-to-face sessions, and in two these were complemented by remote contacts via email or telephone. Interventions

lasted from 1 to 6 months. All studies assessed outcome variables at baseline and post-intervention, and two also included a follow-up measurement to determine whether the changes were maintained at 5 months [40] and 9 months after the intervention [37].

Anderson et al. [38] described assessing adherence to the protocol by recording and analyzing a random sample of visits and telephone calls. This study also included information about the behavior change models and theories on which their intervention was based, specifying the behavioral techniques, such as goal setting, used in the interventions [38] (Table 3).

Table 3. Behavioral techniques and models used in each study.

Study, Country	Behavioral Techniques or Strategies	Behavior Change Models	Measures of Motivation or Barriers
Anderson et al., 2018 [38], UK	Identify what goals mean to participants Realistic goal-setting Implementation intentions	1. Leventhal's self-regulatory theory 2. Social cognitive theory 3. Health action process approach	Beliefs about cancer cause and risk reduction Barriers and motivations
Kiechle et al., 2017 [37], Germany	Not stated	Not stated Mentions theory of planned behavior in a previous publication with the protocol but does not state if the intervention is based on it	N/A
Vrieling et al., 2018 [40], The Netherlands	N/A	Not stated Mentions behavior change theories and models in the Discussion	N/A
Bruno et al., 2020 [39], Italy	Not stated	Not stated in the study or previously published protocol	N/A

Studies were led and monitored by healthcare professionals from diverse backgrounds, including nurses, genetic counsellors, and others. All recruited participants who were attending a genetic counseling unit, but only one study indicated that genetic counsellors led the intervention [40]. One study specifically mentioned that the intervention was nurse-led [38]; the rest did not specify which professionals delivered it [37,39].

3.4. Outcomes

Table 4 details the results of included studies according to the behaviors targeted.

Table 4. Summary of findings across targeted behaviors or risk factors.

Study	Physical Activity	Dietary Intake	Weight/BMI	Alcohol/Tobacco	Other
Anderson et al. [38]	Increase in moderate exercise: 58.1 to 86.8 min in intervention group, 60.3 to 73.2 min in control; no significant difference in vigorous activity	Decrease in dietary fat scores (mean difference −7.8 in intervention group, −1.2 control) Change in fiber intake: +0.6 intervention group, −0.8 control	Mean weight loss: −3.2 kg intervention group, −0.3 kg control	Not reported	Barriers to change: daily routines, sedentary occupations, family commitments, poor physical or mental health, stressful events, complex relationships with food

Table 4. *Cont.*

Study	Physical Activity	Dietary Intake	Weight/BMI	Alcohol/Tobacco	Other
Kiechle et al. [37]	VO_2 peak improved in the intervention group at 3 months, but these effects diminished at 12 months. Aerobic capacity and min of exercise per week did not improve	No differences in the total daily calorie intake or fat intake in either group. Baseline median MEDAS score was 2 points higher in the intervention group versus control ($p = 0.020$); this difference widened significantly at 3 months ($p = 0.001$).	No significant differences between groups	N/A	Women with chronic stress were probably included. At 12 months, median scores on the Short Screening Scale for Chronic Stress were significantly lower in the intervention group compared to control (14.6 versus 20.9; $p = 0.022$). Health-related quality of life was similar between groups.
Vrieling et al. [40]	Adherence to physical activity recommendations improved in both groups	Adherence to the WCRF/AICR recommendations did not differ between groups. Highest adherence rates were found for intake of alcohol and sugary drinks.	No significant differences between groups	Not reported	Awareness and knowledge of the WCRF/AICR recommendations varied by recommendation but were significantly higher in the intervention group compared with the control group for all recommendations.
Bruno et al. [39]	N/A	Compared to control, the intervention group showed significantly increased intake of whole grain products ($p < 0.001$) and legumes, nuts, and seeds ($p = 0.02$), and reduced intake of dairy products ($p = 0.01$) and red and processed meat ($p = 0.04$)	More weight loss ($p < 0.001$) and lower BMI ($p < 0.001$) in intervention vs. control	N/A	Intervention group showed larger reduction in waist circumference ($p = 0.01$), hip circumference ($p = 0.01$), total cholesterol ($p = 0.04$), triglycerides ($p = 0.01$), and IGF-I levels ($p = 0.02$) compared to control.

N/A: not applicable, AICR: American Institute for Cancer Research, WCRF: World Cancer Research Fund, IGF-I: insulin-like growth factor-I.

3.4.1. Diet

Dietary behavior was measured using different questionnaires; two studies [37,39] used the Mediterranean Diet Adherence Screener (MEDAS) [41], and three used different self-reported questionnaires about adherence to recommendations [40] or sections of these [38,39]. Despite differences in data collection methods, the results had commonalities across the studies, with reports of increased fruit and vegetable intake, and, where measured, an increase in fiber and a reduction in red meat intake. Dietary behaviors improved at the post-intervention time point, but the magnitude of the effect showed a sensible decline on follow-up measurements, although they remained better than baseline levels [37,38,40].

3.4.2. Physical Activity

Three studies assessed self-reported physical activity, with two reporting that participants performed more minutes of moderate physical activity at the post-intervention time

point [38]. Another study objectively measured physical activity with monitors such as pedometers [38]. Kiechle et al. [37] chose to measure aerobic capacity (VO_2) as an objective measure of resistance to physical activity.

All studies showed an increase in physical activity, using different outcomes. The post-intervention assessment showed that people increased their physical activity; however, participants who were assessed over follow-up tended to regress towards baseline levels [37].

3.4.3. Weight/Body Mass Index (BMI)

Results on weight and BMI differed between studies; two studies did not report differences between the intervention and control group [37,40], while two studies reported more weight loss and lower BMI in the intervention group [38,39].

3.4.4. Alcohol and Tobacco

Two studies mentioned alcohol [38,40] but did not report outcomes from the intervention. One study also included tobacco among the targets of the intervention [38], but there was no mention of measures or changes.

3.4.5. Motivations and Barriers

The included studies did not assess factors such as motivation for change, readiness for change, or patients' mental health. The feasibility studies showed good motivation and satisfaction with the intervention but reported barriers to adherence, such as duration, travel needs, and personal barriers [37,38]. Some participants also dropped out due to family commitments [39] or lack of motivation [37].

4. Discussion

The findings of this systematic review suggest that healthcare interventions can be useful to modify lifestyle behaviors in adults with a high risk of cancer. However, current evidence is scarce and highly skewed towards interventions for people with a personal history of cancer. Behavioral modifications were not the primary objective of the studies, half of which assessed other parameters (blood test results, awareness of recommendations, acceptability of the intervention) as primary outcomes.

While there is extensive evidence supporting the relationship between modifiable lifestyle behaviors and cancer [3,42], included studies assessed only a few behavioral factors, mainly diet and physical activity. Alcohol intake is associated with the risk of breast, colorectal, liver, and other cancer [43], but it was not addressed in the included articles. This was also the case for tobacco use.

The included studies showed that lifestyle interventions are effective in driving people to increase their physical activity and improve their diet, but these improvements are not sustained over time, regressing towards baseline or remaining slightly better in the case of dietary modification. The findings were not conclusive for weight/BMI changes, as some studies showed no changes and others a greater weight loss in the intervention group.

This review reveals evidence gaps around behavioral interventions in people with a high risk of cancer, especially in those without a personal history of the disease. All studies included some cancer survivors, except Anderson et al., 2018 [38], who included only healthy participants with a family history of cancer. Evidence suggests that people with a confirmed or suspected high risk of cancer do not take the initiative in seeking behavioral recommendations from healthcare professionals [39]. Patients are often unaware or have incorrect perceptions about behavior and cancer risk [19]. In a study that took place in a breast screening service, alcohol was identified as a risk factor by only 19.5% of healthy women attending the screening and by less than half of the healthcare professionals working there [12]. In the study by Vrieling et al. [40], knowledge of cancer risk factors was significantly greater in the intervention versus the control group. Risk perception influences behavior [44], so the lack of awareness of the importance of behavioral factors

among both patients and healthcare professionals is concerning, undermining effective behavior change.

The studies included in this systematic review involved participants who were already motivated (i.e., they agreed to join studies focused on modification of lifestyle behaviors) but still identified some barriers to change. However, none of the included studies addressed these barriers or explored the participants' motivations. Because behavior change is a complicated process [45], designing a behavioral intervention requires understanding the behavior and identifying the intervention options, including the individual's motivation and capacity for change (a process well defined in the Behavior Change Wheel [46]), which the included studies failed to do. One study [38] mentions the behavior change models and the reasoning underpinning the intervention, but none mentioned previous studies supporting the intervention options in relation to behavior change.

The failure to use behavioral techniques or models may also explain the heterogeneity of behavioral interventions in the included studies used leaflets, activities, and consultations. Systematic reviews and studies have compared face-to-face, online, and blended interventions, without a clear preference [47–49]. While the included studies showed some evidence of effecting behavioral changes or even improving awareness and knowledge of cancer risk factors (such as the study by Vrieling et al. [40]), they failed to plan the interventions using models to sustain these changes. The use of behavior change methods and theories to design interventions, together with public and patient involvement during the design and implementation phases, are of critical importance in healthcare interventions. In the context of hereditary cancer, such approaches have proven useful in improving health-seeking behaviors associated with the detection of Lynch syndrome [50].

Patients with a high risk of cancer live with the extra psychological burden of uncertainty about when and if they will develop a cancer [51]. They may be motivated to change, but the goal of reducing cancer risk is not sufficient to maintain these changes over time. Promoting self-care and good mental health favors sustainability [52]. Healthcare professionals should consider these complex needs and build their skills in behavioral interventions to support this population.

Whether behavioral interventions in people at high risk of cancer have a true impact on cancer incidence remains unknown. Just two studies assessed outcomes at follow-up, making it difficult to assess effectiveness over time.

Although genetic counsellors do meet with people at high risk of cancer, they have little opportunity to discuss behavioral factors, as they normally see their patients only twice [18]. Ideally, all health professionals (i.e., genetic counsellors, oncologists, nurses) should be involved in care for people with a high risk of cancer, initiating conversations about lifestyle behaviors and offering evidence-based recommendations on behavioral modifications, also considering the psychosocial support needed to achieve them. This review found different healthcare professionals involved in delivering the interventions, suggesting that there may not be any specific healthcare professionals with the responsibility for addressing lifestyle behaviors in people with inherited cancer syndromes and raising the need to define competencies in that regard [53,54].

Strengths and Limitations

This review included four RCTs of medium or low quality. Due to the limited number of studies in this field, no studies were excluded due to quality, but the quality of the included studies constitutes a limitation.

A further limitation is the heterogeneity of methods and interventions; each study measured different behavioral outcomes in different ways, precluding meta-analysis. Results were instead combined in a narrative synthesis and a table.

The main weakness of the studies was insufficient blinding in participants and unclear blinding in outcome assessors. However, blinding patients in this kind of interventions is not possible, as they have to actively participate in the intervention, so they know which group they belong to. This could also influence their answers on the self-assessment

questionnaires. However, this limitation is not inherent to researchers performing the pre- and post-intervention assessments, who could more easily be blinded.

Including people both with and without a history of cancer poses a limitation. Interventions may have different effects in people with a family history of cancer or an inherited cancer syndrome compared to people with a personal history of cancer, who generally make more lifestyle behavior changes and whose motivation is more about improving quality of life than reducing risks [24]. The studies that included both people with and without a personal history of cancer did not separate or compare the results between populations, precluding any differentiated analysis in our study.

Another limitation is that the results are based on experimental contexts, making the conclusions of the studies more difficult to extrapolate to general practice, as all interventions had specific funding that may be difficult to sustain on finishing the study.

The main strengths of this review were that the included studies were adequately randomized, had comparable groups, and used similar interventions. Added strengths include the review's clear protocol, methods, and the inclusion of six different databases in the search. A variety of studies were included in the review, providing a broad overview of the types of behavioral interventions applied to reduce cancer risk. The review also identified gaps in knowledge and highlighted areas for future research.

5. Conclusions

There are few studies of behavioral interventions for people with a high risk of cancer, and most of these are focused on diet and exercise. The interventions explored showed that behavioral interventions promote positive short-term results, but they fail to promote long-term lifestyle modification.

Future research and interventions should focus on healthcare professionals' knowledge of the impact of behavior on cancer risk, as well as behavior change techniques and promotion of mental health. Strengthening the competencies of healthcare professionals in this regard can help in identifying and supporting the needs of people at high risk of cancer.

Supplementary Materials: The following supporting information can be downloaded at: https://www.mdpi.com/article/10.3390/ijerph192114098/s1, Table S1: PRISMA Checklist of items to include when reporting a systematic review or meta-analysis [29]; Table S2: Free-text and MeSH terms pertinent to three strings of search. String A was first combined with string B, and subsequently with string C; Table S3: Example of PubMed search.

Author Contributions: Conceptualization, C.D.d.l.R.d.l.S., P.F.-O., and T.L.-C.; methodology, C.D.d.l.R.d.l.S., P.F.-O., and T.L.-C.; validation, C.D.d.l.R.d.l.S., P.F.-O., and T.L.-C.; formal analysis, C.D.d.l.R.d.l.S., P.F.-O., and T.L.-C.; investigation, C.D.d.l.R.d.l.S.; writing—original draft preparation, C.D.d.l.R.d.l.S.; writing—review and editing, C.D.d.l.R.d.l.S., P.F.-O. and T.L.-C.; visualization, C.D.d.l.R.d.l.S.; supervision, P.F.-O. and T.L.-C. All authors have read and agreed to the published version of the manuscript.

Funding: This research received no external funding.

Institutional Review Board Statement: Not applicable.

Informed Consent Statement: Not applicable

Data Availability Statement: The review was registered on PROSPERO: CRD42020209921.

Acknowledgments: We would like to thank UB librarians for their support in the literature search, Greg Kotronoulas and Vanessa Seilo for their expert assistance improving this article prior submission.

Conflicts of Interest: The authors declare no conflict of interest.

References

1. Wu, S.; Zhu, W.; Thompson, P.; Hannun, Y.A. Evaluating intrinsic and non-intrinsic cancer risk factors. *Nat. Commun.* **2018**, *9*, 3490. [CrossRef] [PubMed]
2. Ferlay, J.; Colombet, M.; Soerjomataram, I.; Mathers, C.; Parkin, D.M.; Piñeros, M.; Znaor, A.; Bray, F. Estimating the global cancer incidence and mortality in 2018: GLOBOCAN sources and methods. *Int. J. Cancer* **2019**, *144*, 1941–1953. [CrossRef] [PubMed]
3. Whiteman, D.C.; Wilson, L.F. The fractions of cancer attributable to modifiable factors: A global review. *Cancer Epidemiol.* **2016**, *44*, 203–221. [CrossRef]
4. Marzo-Castillejo, M.; Vela-Vallespín, C.; Bellas-Beceiro, B.; Bartolomé-Moreno, C.; Melús-Palazón, E.; Vilarrubí-Estrella, M.; Nuin-Villanueva, M. Recomendaciones de prevención del cáncer. Actualización PAPPS 2018. *Aten. Primaria.* **2018**, *50*, 41–65. [CrossRef]
5. Foulkes, W.D. Inherited Susceptibility to Common Cancers. *N. Engl. J. Med.* **2008**, *359*, 2143–2153. [CrossRef] [PubMed]
6. Zhu, M.; Wang, T.; Huang, Y.; Zhao, X.; Ding, Y.; Zhu, M.; Ji, M.; Wang, C.; Dai, J.; Yin, R.; et al. Genetic Risk for Overall Cancer and the Benefit of Adherence to a Healthy Lifestyle. *Cancer Res.* **2021**, *81*, 4618–4627. [CrossRef] [PubMed]
7. Lazzeroni, M.; Bellerba, F.; Calvello, M.; Macrae, F.; Win, A.; Jenkins, M.; Serrano, D.; Marabelli, M.; Cagnacci, S.; Tolva, G.; et al. A Meta-Analysis of Obesity and Risk of Colorectal Cancer in Patients with Lynch Syndrome: The Impact of Sex and Genetics. *Nutrients* **2021**, *13*, 1736. [CrossRef]
8. Daniele, A.; Divella, R.; Pilato, B.; Tommasi, S.; Pasanisi, P.; Patruno, M.; Digennaro, M.; Minoia, C.; Dellino, M.; Pisconti, S.; et al. Can harmful lifestyle, obesity and weight changes increase the risk of breast cancer in BRCA 1 and BRCA 2 mutation carriers? A Mini review. *Hered. Cancer Clin. Pract.* **2021**, *19*, 45. [CrossRef]
9. Coletta, A.M.; Peterson, S.K.; Gatus, L.A.; Krause, K.J.; Schembre, S.M.; Gilchrist, S.C.; Arun, B.; You, Y.N.; Rodriguez-Bigas, M.A.; Strong, L.L.; et al. Diet, weight management, physical activity and Ovarian & Breast Cancer Risk in women with BRCA1/2 pathogenic Germline gene variants: Systematic review. *Hered. Cancer Clin. Pract.* **2020**, *18*, 5.
10. Carr, P.R.; Weigl, K.; Edelmann, D.; Jansen, L.; Chang-Claude, J.; Brenner, H.; Hoffmeister, M. Estimation of Absolute Risk of Colorectal Cancer Based on Healthy Lifestyle, Genetic Risk, and Colonoscopy Status in a Population-Based Study. *Gastroenterology* **2020**, *159*, 129–138. [CrossRef]
11. Wild, C.P.; Espina, C.; Bauld, L.; Bonanni, B.; Brenner, H.; Brown, K.; Dillner, J.; Forman, D.; Kampman, E.; Nilbert, M.; et al. Cancer Prevention Europe. *Mol. Oncol.* **2019**, *13*, 528–534. [CrossRef]
12. Sinclair, J.; McCann, M.; Sheldon, E.; Gordon, I.; Brierley-Jones, L.; Copson, E. The acceptability of addressing alcohol consumption as a modifiable risk factor for breast cancer: A mixed method study within breast screening services and symptomatic breast clinics. *BMJ Open.* **2019**, *9*, e027371. [CrossRef] [PubMed]
13. World Cancer Research Fund International. *Our Cancer Prevention Recommendations*; WCRF: London, UK, 2016; Available online: https://www.wcrf.org/diet-and-cancer/cancer-prevention-recommendations/ (accessed on 22 August 2022).
14. Makarem, N.; Lin, Y.; Bandera, E.V.; Jacques, P.F.; Parekh, N. Concordance with World Cancer Research Fund/American Institute for Cancer Research (WCRF/AICR) guidelines for cancer prevention and obesity-related cancer risk in the Framingham Offspring cohort (1991–2008). *Cancer Causes Control* **2015**, *26*, 277–286. [CrossRef]
15. Schüz, J.; Espina, C.; Villain, P.; Herrero, R.; Leon, M.E.; Minozzi, S.; Romieu, I.; Segnan, N.; Wardle, J.; Wiseman, M.; et al. European Code against Cancer 4th Edition: 12 ways to reduce your cancer risk. *Cancer Epidemiol.* **2015**, *39*, S1–S10. [CrossRef]
16. McGuinness, L.A.; Higgins, J.P.T. Risk-of-bias VISualization (robvis): An R package and Shiny web app for visualizing risk-of-bias assessments. *Res. Synth. Methods* **2021**, *12*, 55–61. [CrossRef]
17. Abacan, M.A.; Alsubaie, L.; Barlow-Stewart, K.; Caanen, B.; Cordier, C.; Courtney, E.; Davoine, E.; Edwards, J.; Elackatt, N.J.; Gardiner, K.; et al. The Global State of the Genetic Counseling Profession. *Eur. J. Hum. Genet.* **2019**, *27*, 183–197. [CrossRef]
18. Resta, R.G. What have we been trying to do and have we been any good at it? A history of measuring the success of genetic counseling. *Eur. J. Med. Genet.* **2019**, *62*, 300–307. [CrossRef] [PubMed]
19. Quillin, J.M. Lifestyle Risk Factors Among People Who Have Had Cancer Genetic Testing. *J. Genet. Couns.* **2016**, *25*, 957–964. [CrossRef]
20. Lammert, J.; Grill, S.; Kiechle, M. Modifiable Lifestyle Factors: Opportunities for (Hereditary) Breast Cancer Prevention—A Narrative Review. *Breast Care.* **2018**, *13*, 109–114. [CrossRef]
21. Nippert, I.; Julian-Reynier, C.; Harris, H.; Evans, G.; Van Asperen, C.J.; Tibben, A.; Schmidtke, J. Cancer risk communication, predictive testing and management in France, Germany, the Netherlands and the UK: General practitioners' and breast surgeons' current practice and preferred practice responsibilities. *J. Community Genet.* **2014**, *5*, 69–79. [CrossRef]
22. Hollands, G.J.; French, D.P.; Griffin, S.J.; Prevost, A.T.; Sutton, S.; King, S.; Marteau, T.M. The impact of communicating genetic risks of disease on risk reducing health behaviour: Systematic review with meta-analysis. *BMJ* **2016**, *352*, i1102. [CrossRef]
23. Orange, S.T.; Hicks, K.M.; Saxton, J.M. Effectiveness of diet and physical activity interventions amongst adults attending colorectal and breast cancer screening: A systematic review and meta-analysis. *Cancer Causes Control* **2021**, *32*, 13–26. [CrossRef]
24. Grimmett, C.; Simon, A.; Lawson, V.; Wardle, J. Diet and physical activity intervention in colorectal cancer survivors: A feasibility study. *Eur. J. Oncol. Nurs.* **2015**, *19*, 1–6. [CrossRef]
25. Spees, C.K.; Braun, A.C.; Hill, E.B.; Grainger, E.M.; Portner, J.; Young, G.S.; Kleinhenz, M.D.; Chitchumroonchokchai, C.; Clinton, S.K. Impact of a Tailored Nutrition and Lifestyle Intervention for Overweight Cancer Survivors on Dietary Patterns, Physical Activity, Quality of Life, and Cardiometabolic Profiles. *J. Oncol.* **2019**, *2019*, 1503195. [CrossRef]

26. Michie, S.; Atkins, L.; West, R. *The Behaviour Change Wheel. A Guide to Designing Interventions*, 1st ed.; Silverback Publishing: Sutton, UK, 2016; pp. 1003–1010.
27. Edwards, A.G.K.; Naik, G.; Ahmed, H.; Elwyn, G.J.; Pickles, T.; Hood, K.; Playle, R. Personalised risk communication for informed decision making about taking screening tests. *Cochrane Database Syst. Rev.* **2013**, 1–96. [CrossRef] [PubMed]
28. Tufanaru, C.; Munn, Z.; Aromataris, E.; Campbell, J.; Hopp, L. JBI Manual for Evidence Synthesis. In *JBI Manual for Evidence Synthesis*; JBI: Adelaide, Australia, 2020.
29. Moher, D.; Liberati, A.; Tetzlaff, J.; Altman, D.G. *Preferred Reporting Items for Systematic Reviews and Meta-Analyses: The PRISMA Statement*; BMJ (Online); British Medical Journal Publishing Group: London, UK, 2009; Volume 339, pp. 332–336. Available online: http://www.bmj.com/content/339/bmj.b2535?tab=related#datasupp (accessed on 13 September 2020).
30. Centre for Reviews and Dissemination; Akers, J. *Systematic Reviews: CRD's Guidance for Undertaking Reviews in Health Care*; CRD, University of York: York, UK, 2009; Available online: www.york.ac.uk/inst/crd (accessed on 13 September 2022).
31. Underhill-Blazey, M.; Rodriguez, D.; Norton, S.A. Scoping Review of Nonsurgical, Nonpharmacologic Interventions After Risk Reduction: Improving Quality of Life for Patients with Inherited Cancer Risk. *Oncol. Nurs. Forum* **2022**, *49*, 193–200. [CrossRef]
32. Malakou, E.; Linardakis, M.; Armstrong, M.E.G.; Zannidi, D.; Foster, C.; Johnson, L.; Papadaki, A. The Combined Effect of Promoting the Mediterranean Diet and Physical Activity on Metabolic Risk Factors in Adults: A Systematic Review and Meta-Analysis of Randomised Controlled Trials. *Nutrients* **2018**, *10*, 1577. [CrossRef] [PubMed]
33. André, N.; Pillaud, M.; Davoust, A.; Laurencelle, L. Barriers Identification as Intervention to Engage Breast Cancer Survivors in Physical Activity. *Psychosoc. Interv.* **2018**, *27*, 35–43. [CrossRef]
34. Sterne, J.A.C.; Savović, J.; Page, M.J.; Elbers, R.G.; Blencowe, N.S.; Boutron, I.; Cates, C.J.; Cheng, H.Y.; Corbett, M.S.; Eldridge, S.M.; et al. RoB 2: A revised tool for assessing risk of bias in randomised trials. *BMJ* **2019**, *366*, l4898. [CrossRef]
35. Tufanaru, C.; Munn, Z.; Aromataris, E.; Campbell, J.; Hopp, L. Chapter 3: Systematic Reviews of Effectiveness. *JBI Man. Evid. Synth.* 2020. Available online: https://synthesismanual.jbi.global (accessed on 7 September 2022).
36. Hoffmann, T.C.; Glasziou, P.P.; Boutron, I.; Milne, R.; Perera, R.; Moher, D.; Altman, D.G.; Barbour, V.; Macdonald, H.; Johnston, M.; et al. Better reporting of interventions: Template for intervention description and replication (TIDieR) checklist and guide. *BMJ* **2014**, *348*, g1687. [CrossRef]
37. Kiechle, M.; Dukatz, R.; Yahiaoui-Doktor, M.; Berling, A.; Basrai, M.; Staiger, V.; Niederberger, U.; Marter, N.; Lammert, J.; Grill, S.; et al. Feasibility of structured endurance training and Mediterranean diet in BRCA1 and BRCA2 mutation carriers—An interventional randomized controlled multicenter trial (LIBRE-1). *BMC Cancer* **2017**, *17*, 752. [CrossRef] [PubMed]
38. Anderson, A.S.; Dunlop, J.; Gallant, S.; Macleod, M.; Miedzybrodzka, Z.; Mutrie, N.; O'Carroll, R.E.; Stead, M.; Steele, R.J.; Taylor, R.S.; et al. Feasibility study to assess the impact of a lifestyle intervention (a € LivingWELL') in people having an assessment of their family history of colorectal or breast cancer. *BMJ Open.* **2018**, *8*, e019410. [CrossRef] [PubMed]
39. Bruno, E.; Oliverio, A.; Paradiso, A.V.; Daniele, A.; Tommasi, S.; Tufaro, A.; Terribile, D.A.; Magno, S.; Filippone, A.; Venturelli, E.; et al. A Mediterranean Dietary Intervention in Female Carriers of BRCA Mutations: Results from an Italian Prospective Randomized Controlled Trial. *Cancers* **2020**, *12*, 3732. [CrossRef]
40. Vrieling, A.; Visser, A.; Hoedjes, M.; Hurks, M.; Gómez García, E.; Hoogerbrugge, N.; Kampman, E. Increasing awareness and knowledge of lifestyle recommendations for cancer prevention in Lynch syndrome carriers: Randomized controlled trial. *Clin. Genet.* **2018**, *93*, 67–77. [CrossRef]
41. Schröder, H.; Fitó, M.; Estruch, R.; Martínez-González, M.A.; Corella, D.; Salas-Salvadó, J.; Lamuela-Raventós, R.; Ros, E.; Salaverría, I.; Fiol, M.; et al. A Short Screener Is Valid for Assessing Mediterranean Diet Adherence among Older Spanish Men and Women. *J. Nutr.* **2011**, *141*, 1140–1145. [CrossRef] [PubMed]
42. Fujiyoshi, K.; Sudo, T.; Fujita, F.; Chino, A.; Akagi, K.; Takao, A.; Yamada, M.; Tanakaya, K.; Ishida, H.; Komori, K.; et al. Risk of First Onset of Colorectal Cancer Associated with Alcohol Consumption in Lynch Syndrome: A Multicenter Cohort Study. *Int. J. Clin. Oncol.* **2022**, *27*, 1051–1059. [CrossRef] [PubMed]
43. Papadimitriou, N.; Markozannes, G.; Kanellopoulou, A.; Critselis, E.; Alhardan, S.; Karafousia, V.; Kasimis, J.C.; Katsaraki, C.; Papadopoulou, A.; Zografou, M.; et al. An umbrella review of the evidence associating diet and cancer risk at 11 anatomical sites. *Nat. Commun.* **2021**, *12*, 4579. [CrossRef]
44. Usher-Smith, J.A.; Silarova, B.; Sharp, S.J.; Mills, K.; Griffin, S.J. Effect of interventions incorporating personalised cancer risk information on intentions and behaviour: A systematic review and meta-analysis of randomised controlled trials. *BMJ Open* **2018**, *8*, 17717. [CrossRef]
45. Prochaska, J.O.; Velicer, W.F. The transtheoretical model of health behavior change. *Am. J. Health Promot.* **1997**, *12*, 38–48. [CrossRef]
46. Michie, S.; van Stralen, M.M.; West, R. The behaviour change wheel: A new method for characterising and designing behaviour change interventions. *Implement. Sci.* **2011**, *6*, 42. [CrossRef]
47. Hohberg, V.; Fuchs, R.; Gerber, M.; Künzler, D.; Paganini, S.; Faude, O. Blended Care Interventions to Promote Physical Activity: A Systematic Review of Randomized Controlled Trials. *Sports Med. Open* **2022**, *8*, 100. [CrossRef] [PubMed]
48. Santarossa, S.; Kane, D.; Senn, C.Y.; Woodruff, S.J. Exploring the Role of In-Person Components for Online Health Behavior Change Interventions: Can a Digital Person-to-Person Component Suffice? *J. Med. Internet Res.* **2018**, *20*, e144. [CrossRef] [PubMed]

49. Lavoie, A.; Dubé, V. Web-Based Interventions to Promote Healthy Lifestyles for Older Adults: Scoping Review. *Interact. J. Med. Res.* **2022**, *11*, e37315. [CrossRef]
50. Long, J.C.; Winata, T.; Debono, D.; Phan-Thien, K.C.; Zhu, C.; Taylor, N. Process evaluation of a behaviour change approach to improving clinical practice for detecting hereditary cancer. *BMC Health Serv. Res.* **2019**, *19*, 180. [CrossRef] [PubMed]
51. Spencer, J.C.; Wheeler, S.B. A systematic review of Motivational Interviewing interventions in cancer patients and survivors. *Patient Educ. Couns.* **2016**, *99*, 1099–1105. [CrossRef]
52. Teixeira, S.M.A.; Coelho, J.C.F.; Sequeira, C.A.D.C.; Lluch i Canut, M.T.; Ferre-Grau, C. The effectiveness of positive mental health programs in adults: A systematic review. *Health Soc. Care Community* **2019**, *27*, 1126–1134. [CrossRef] [PubMed]
53. Chambers, S.E.; Copson, E.R.; Dutey-Magni, P.F.; Priest, C.; Anderson, A.S.; Sinclair, J.M.A. Alcohol use and breast cancer risk: A qualitative study of women's perspectives to inform the development of a preventative intervention in breast clinics. *Eur. J. Cancer Care (Engl).* **2019**, *28*, e13075. [CrossRef] [PubMed]
54. Anderson, A.S.; Caswell, S.; Macleod, M.; Steele, R.J.; Berg, J.; Dunlop, J.; Stead, M.; Eadie, D.; O'carroll, R.E. Health Behaviors and their Relationship with Disease Control in People Attending Genetic Clinics with a Family History of Breast or Colorectal Cancer. *J. Genet. Couns.* **2017**, *26*, 40–51. [CrossRef]

Article 2:

Title	Educational Programme for Cancer Nurses in Genetics, Health Behaviors and Cancer Prevention: A Multidisciplinary Consensus Study
Authors	Diez de los Rios de la Serna, C., Fernández-Ortega, P., Lluch-Canut, T
Reference	Diez de los Rios de la Serna C, Fernández-Ortega P, Lluch-Canut T. Educational Programme for Cancer Nurses in Genetics, Health Behaviors and Cancer Prevention: A Multidisciplinary Consensus Study. J Pers Med. 2022;12(7):1104.
Journal	Journal of Personalized Medicine, 12, 1004
Keywords	Hereditary cancer; hereditary cancer syndromes; genetic testing; nurse education; health behaviors; behavioural change
Impact Factor	3.4
Category	Medicine
Quartile	Q2
ISSN	2075-4426

Journal of *Personalized Medicine*

MDPI

Article

Educational Programme for Cancer Nurses in Genetics, Health Behaviors and Cancer Prevention: A Multidisciplinary Consensus Study

Celia Diez de los Rios de la Serna *,†, Paz Fernández-Ortega and Teresa Lluch-Canut

School of Nursing, Faculty of Medicine and Health Sciences, Bellvitge Campus, Barcelona University, 08036 Barcelona, Spain; mfo@iconcologia.net (P.F.-O.); tlluch@ub.edu (T.L.-C.)
* Correspondence: cdiezdde7@alumnes.ub.edu or celiadiezrios@gmail.com
† PhD candidate.

Abstract: (1) Background: Most common hereditary cancers in Europe have been associated with lifestyle behaviors, and people affected are lacking follow up care. However, access to education programmes to increase knowledge on cancer and genetics and promote healthy lifestyle behaviors in people at high risk of cancer is scarce. This affects the quality of care of people with a hereditary risk of cancer. This study aimed to reach a multidisciplinary consensus on topics and competencies and competencies that cancer nurses need in relation to cancer, genetics, and health promotion. (2) Methods: A two-round online Delphi study was undertaken. Experts in cancer and genetics were asked to assess the relevance of eighteen items and to suggest additional terms. Consensus was defined as an overall agreement of at least 75%. (3) Results: A total of 74 multiprofessional experts from all around the world participated in this study including healthcare professionals working in genetics (39%), researchers in cancer and genetics (31%) and healthcare professionals with cancer patients (30%). Thirteen additional items were proposed. A total of thirty-one items reached consensus. (4) Conclusions: This multidisciplinary consensus study provide the essential elements to build an educational programme to increase cancer nurses' skills to support the complex care of people living with a higher risk of cancer including addressing lifestyle behaviors. All professionals highlighted the importance of cancer nurses increasing their skills in cancer and genetics.

Keywords: hereditary cancer; hereditary cancer syndromes; genetic testing; nurse education; health behaviors; behavioural change

Citation: Diez de los Rios de la Serna, C.; Fernández-Ortega, P.; Lluch-Canut, T. Educational Programme for Cancer Nurses in Genetics, Health Behaviors and Cancer Prevention: A Multidisciplinary Consensus Study. *J. Pers. Med.* **2022**, *12*, 1104. https://doi.org/10.3390/jpm12071104

Academic Editor: Daryl Pritchard

Received: 14 June 2022
Accepted: 1 July 2022
Published: 5 July 2022

1. Introduction

Cancer is the result of genetic changes in the cells that affect cell function [1]. While it can affect anyone, some people are at a higher risk than others. Altogether, about 5% to 10% of cancers are attributable to hereditary genetic alterations [2]. These mutations are infrequent, affecting just over 1% of the population [3], but they considerably increase the risk of developing more than one type of cancer.

Research advances and increased knowledge are paving the way for a greater role of genetics in oncology, offering different possibilities to those with cancer due to a hereditary genetic syndrome. Personalised cancer medicine has become crucial to every stage of cancer care, from prevention to treatment. Next generation sequencing (NGS) using panels of multiple genes that could be linked to cancer risk and the increasingly affordable prices of genetic testing have fostered these changes.

Different authors have investigated whether the population should be routinely screened for syndromes such as hereditary breast and ovarian cancer or Lynch syndrome, as the identification of these syndromes before a cancer diagnosis offers screening and prevention options [4,5]. The detection of genetic mutations also affects cancer treatment

options for the individual and preventive approaches for their family members. For example, BRCA carriers can benefit from the development of targeted treatments such as PARP inhibitors [6], and subsequent to a carrier diagnosis, family testing is recommended. This offers new opportunities for early detection but poses challenges for the healthcare system and the patient in charge of family communication [7].

People suspected of having a hereditary genetic syndrome traditionally see a genetic counsellor, who helps them to understand their genetic risk and decide how to use the new information to adapt their behaviour [8]. In oncology, this activity is focused on patients and their relatives who may be at increased risk of cancer even if they do not currently have a diagnosis. This process involves medical, psychological, and family implications and should include information on therapeutic and preventive decisions along with individual, family, and behavioural risks [9].

However, the increasing role of genetics in oncological treatments poses a challenge to the healthcare system. Families with genetic syndromes expect well-trained professionals to assist in their long-term management, but they often perceive a lack of knowledge in the healthcare professionals performing their follow-up and feel frustration when looking for answers to their questions and worries [10,11]. There is a need to increase the knowledge of oncology professionals on genetics. Experts in cancer and genetics and genetic counsellors from around the world agree that healthcare systems are not prepared to absorb the rising demand for genetic testing. Most also agree that healthcare professionals not working in genetics need to have a role, but the lack of adequate knowledge poses a risk of mismanaging the results [12,13].

For the comprehensive management of people with genetic syndromes, educational needs go further than genetics [11]. Preventive health interventions for people at increased risk of cancer need to focus on health promotion and behaviour, not only on risk reduction strategies such as mastectomies or colonoscopies. Some hospitals have clinical services for people with a family history of cancer, including behavioural counselling and screening, but this is still uncommon [14,15]. Other centres are working to implement mainstream services or specialised follow-up care for families. All these services are delivered mainly by cancer nurses, who play a central role in providing information and empowering patients to take control and participate in their care [16].

However, advances in treatments and rising demands from patients are outpacing progress in training for healthcare professionals. Studies worldwide show that nurses' knowledge about cancer and genetics and hereditary cancer syndromes is low, and they lack confidence when talking about genetics [17–20]. The International Society of Nurses in Genetics (ISONG) highlighted the need to train nurses and build their confidence in talking about genomics in order to better serve their patients [21].

Some countries and professional societies are already studying and incorporating cancer and genetic competencies for nurses, but most competency frameworks are not specific to oncology and do not include competencies on health promotion and lifestyle behaviours [21–23]. While health promotion is normally part of nursing training there is little involvement of cancer nurses in cancer prevention and risk reduction strategies and to empower individuals to take control and participate in their care [24]. The European Oncology Nursing Society developed the EONS Cancer Nursing Education Framework [25], which includes hereditary cancer syndromes, health promotion and risk reduction strategies in its competencies. Access to training on hereditary cancer syndromes is scarce [26] and generally focuses on assessment and testing, not health promotion and follow-up care.

The aim of this study was to reach a consensus on the topics and competencies that cancer nurses need in relation to cancer, genetics, and health promotion. These results can guide the development of a comprehensive educational programme for cancer nurses.

2. Materials and Methods

The study used a Delphi technique [27] to collect professionals' perspective on training needs for cancer nurses related to the topics of genetics, hereditary cancer syndromes, and health promotion.

This investigation is part of a larger project on the role of nurses in health promotion for people with hereditary cancer syndromes. Ethics approval was obtained from the University of Barcelona's Research Ethics Committee (IRB00003099).

2.1. Delphi Study Process

The Delphi method was applied to achieve consensus among a panel of experts about the interventions and knowledge needed to promote healthy behaviours in people at a high risk of cancer [28]. Delphi studies were previously used to plan educational curricula, as using expert clinicians' opinions for this aim is appropriate [29,30]. The technique requires input from experts in the form of anonymised opinions, which are then subjected to a participatory review of the general results that informs the next round, allowing panellists to reflect and revalidate their decisions [31].

A two-round online Delphi survey was performed between February and April 2022. A third round was planned but not needed, as all items reached consensus in the second round. The institution's Microsoft Forms was used to develop the online questionnaire and to keep the information secure on the university servers.

The first step for developing the Delphi survey drew on data from a previous systematic review by the authors of this study to identify what interventions had been administered to patients with a high risk of cancer [32]. There were also items from the module on risk reduction, early detection, and health promotion in cancer care from the EONS Cancer Nursing Education Framework [25]. Items were divided into domains on genetics, behaviours, and communication and barriers, and each included knowledge and skills competencies.

In total, the literature review yielded 18 potential topics based on skills and knowledge competencies; these were incorporated into an online survey. The first round was open for four weeks, and participants were asked to rate the importance of topics for inclusion in the programme using a seven-point Likert scale (1–3 = not important, 4 = unsure, 5–7 = important). In the first round, the questionnaire also contained free text fields where experts could suggest additional competencies not mentioned in the survey.

The survey was first piloted with two experts acquainted with the authors; after incorporating their suggestions, it was sent to the participants [33].

The first round also elicited information about the experts themselves, including country of residence, profession, years of working experience in cancer care, years of working experience in cancer and genetics, and professional roles. Their email was requested in order to contact them for the next rounds.

Data were exported to an Excel spreadsheet, anonymised by one investigator (C.D.R.S.), and analysed by all authors (C.D.R.S., P.F.-O., T.L.-C.). The authors undertook a descriptive analysis of the Likert scale, measuring the percentage of agreement about which items were important, not important, or of uncertain importance. As with the size of the panel, there is no general definition for what constitutes consensus in Delphi studies, but a threshold of 60% or more is typical; for this study, consensus was defined as agreement of 75% or more [31]. Content analysis was used to evaluate comments and suggest additional items. Items were grouped into themes and were either incorporated into existing competencies when appropriate or defined as new competencies by author agreement.

In the second round, experts had access to the previous round of results. They gave their opinion on all the first-round items as well as on the new items generated from the suggestions in the previous round. Reminders were sent to all the experts from the first round after two weeks. This round also had a final free text box for comments. All the final results and comments were shared with the participants.

2.2. Expert Panel

All participants were health professionals from different European countries with expertise in cancer and genetics. Invitations were sent to different members of multidisciplinary care teams with expertise in high-risk cancer patients, including oncologists, cancer nurses, genetic counsellors, nutritionists, physical therapists, psychologists, and other professionals involved in care for these patients. In addition, researchers and other people with specific expertise in the field were invited, such as authors of papers relevant to the topic and members of the European Society of Human Genetics, Cancer Genetics Group, the International Society of Nurses in Genetics, and the Global Genomics Nursing Alliance group. The study was also advertised on Twitter to identify possible participants. Other experts joined the panel through snowball sampling, as panellists were encouraged to invite other experts. Participants that expressed interest received a link to the online survey.

There is no defined number for a Delphi study panel, but as it depends on a group of experts to reach consensus, a panel of around 18 experts is recommended to ensure sufficient contributions. Assuming an attrition rate of 50%, this study planned to recruit a minimum of 36 participants [33,34].

3. Results

The Delphi study took place between January and April 2022. Details of the Delphi process can be seen in Figure 1.

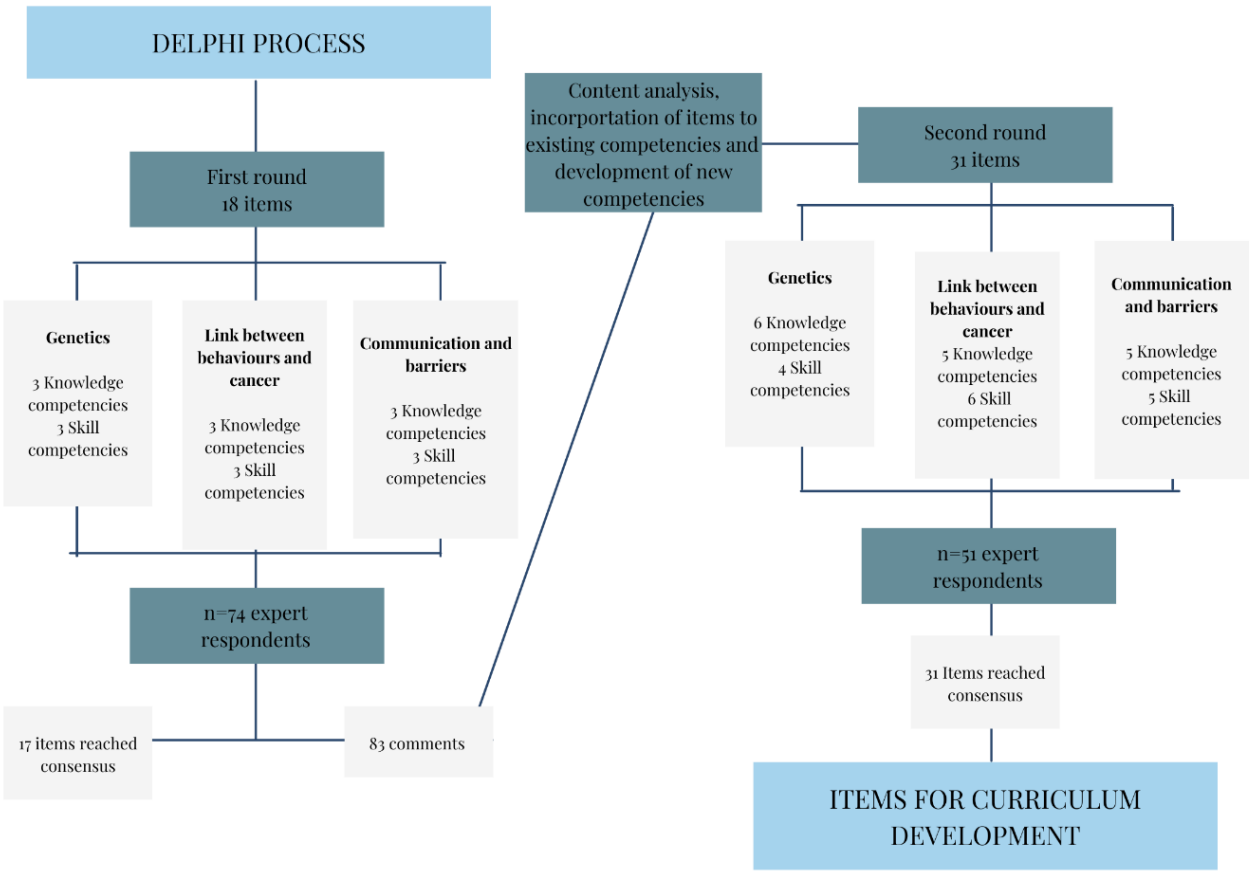

Figure 1. Flow chart of the Delphi process.

Expert Characteristics

In the first round, 74 experts participated in the Delphi study. All panellists were healthcare professionals from different backgrounds and professions with expertise in both cancer and genetics. Thirty-nine per cent were healthcare professionals working in genetics, 31% were healthcare professionals looking after cancer patients, and 30% were

researchers in cancer and genetics (Figure 2a). Panellists were nurses (n = 30), physicians (n = 20), genetic counsellors (n = 9), academics/researchers (n = 10), nutritionists (n = 3), and psychologists (n = 2). Forty-four per cent had more than 20 years of experience (Figure 2b).

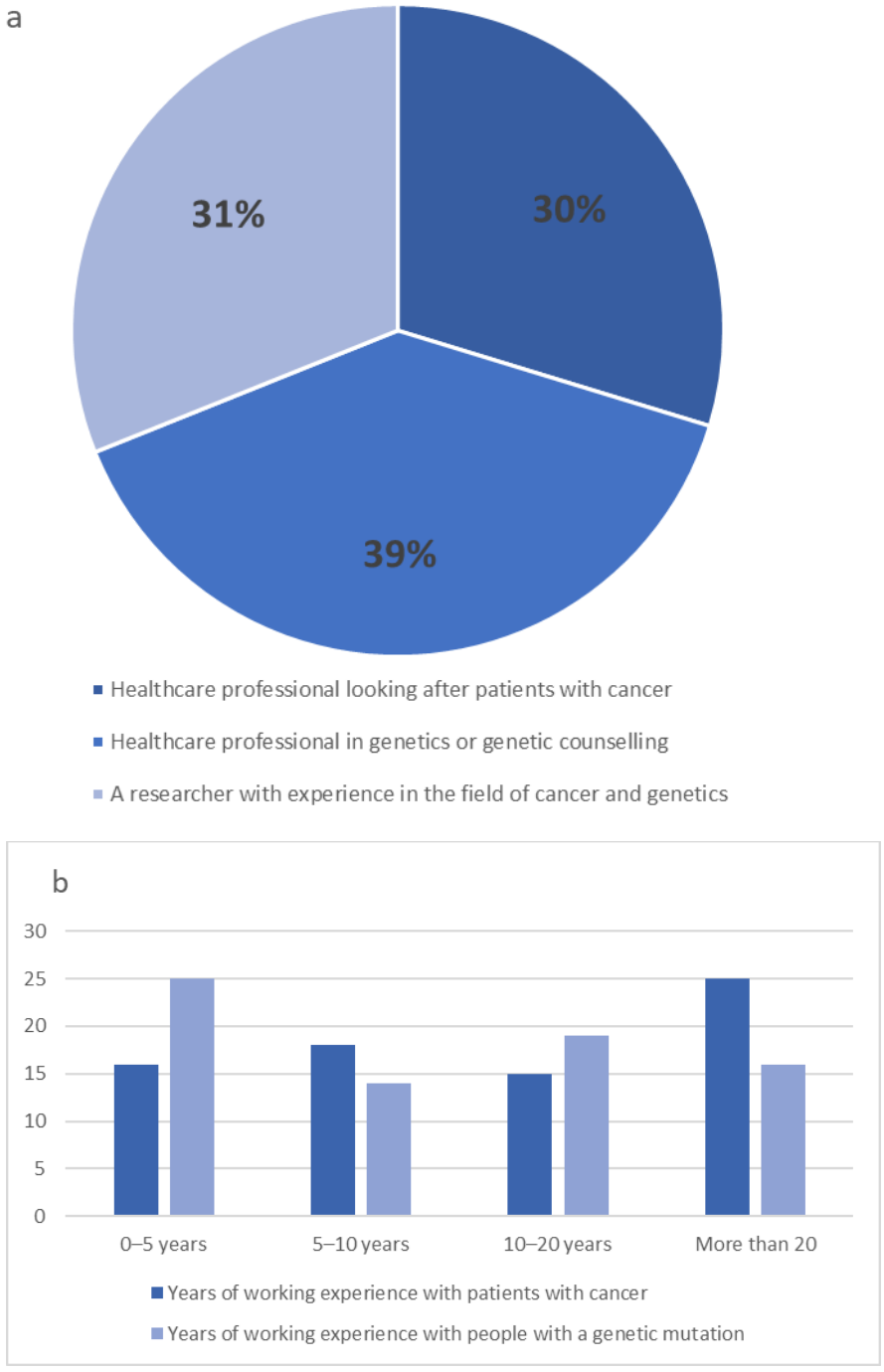

Figure 2. Description of the Delphi participants (**a**) Clinical experience of the Delphi participants (**b**) Years of experience of the Delphi participants.

Participants were predominantly from Europe (n = 61), but there were also three participants from Hong Kong, five from the USA, two from Australia, one from Israel, one from New Zealand and one from Japan. Of the 82.4% of European experts, most were from the UK (n = 13) and Spain (n = 11). There were also participants from Estonia (n = 5), Ireland (n = 5), the Netherlands (n = 4), Sweden (n = 3), Portugal (n = 3), Finland (n = 2),

France (n = 2), and Serbia (n = 2), as well as one each from Belgium, Croatia, Czechia, Denmark, Greece, Italy, Latvia, Norway, Slovenia, Switzerland, and Turkey (Figure 3).

Figure 3. Map of European participants n = number of participants from each European country.

In the first round, the panel reached a consensus on 17 of the 18 items proposed being classified as important. The remaining competency, "Knowledge on the European Code Against Cancer recommendations", was important for just 68% of the panellists, and some people reported not being aware of the code or thinking there could be many other recommendations, such as the World Cancer Research Fund recommendations. Experts also provided 87 comments that were subsequently analysed.

Some of the suggestions were similar and therefore grouped together; for example, three panellists suggested skills for drawing pedigrees; five on basic knowledge on counselling; and four on competencies for follow-up and support after diagnosis with genetic counselling. The 87 comments were consolidated into 42 items. Then, those that could be encompassed under existing competencies were added with an explicit specification so participants in round 2 could see how their comments were used to modify existing items or create new ones.

The second round listed 31 items in the following two groups: existing items where participants could see results from round 1 and new items based on panellist proposals. The round 2 survey also had an option to provide free text comments at the end.

Fifty-one (68.9%) participants from the first round completed the round 2 survey. All the items were considered important by enough participants to reach consensus. The degree of consensus was 80% or higher for most items, while only the following two items had a lower level of agreement, at 76%: "Knowledge of recommendations to reduce risk of cancer (the item previously called "Knowledge on the European Code Against Cancer recommendations") and "Knowledge on health belief theories and health behaviour change theories", which yielded a 78% consensus during the first round (Table 1).

Table 1. Educational items proposed in the two Delphi rounds and % of agreement.

Genetics	Round 1 Agreement	Round 2 Agreement
Knowledge on determinants of cancer	100%	100%
Knowledge and understanding of the most common genetic mutations/syndromes in cancer setting	95%	95%
Knowledge of the role of genetics in cancer treatment	93%	95%
Knowledge of instruments to estimate risk	NEW in 2	80%
Knowledge on genetic processes	NEW in 2	88%
Knowledge of the role of genetic counsellors	NEW in 2	80%
Ability to undertake a comprehensive history to identify the individual, familial, genetic, sociocultural, economic and environmental factors	93%	96%
Ability to identify individuals that may be potentially at risk of having a genetic predisposition to cancer	97%	96%
Ability to create communication links between oncology and genetic healthcare providers	96%	92%
Ability to explain patients genetic testing	NEW in 2	84%
Behaviors	**Round 1**	**Round 2**
Knowledge on modifiable determinants of cancer and their importance on people with high risk of cancer	92%	90%
Health promotion and health education	91%	84%
Knowledge on the European Code Against Cancer recommendations changed in the second round to: Knowledge of recommendations to reduce risk of cancer	64%	76%
Knowledge of the social and behavioural determinants of health on genetic susceptibility	NEW in 2	86%
Surveillance	NEW in 2	88%
Ability to use health promotion/disease prevention practices that incorporate genetic and genomic information as well as personal and environmental risk factors	91%	88%
Ability to address peoples' beliefs and values	92%	94%
Ability to identify problems with surveillance	86%	92%
Ability to recognise risk factors	NEW in 2	80%
Communication	**Round 1**	**Round 2**
Barriers to effective information provision	92%	93%
Awareness of consequences of cancer such as the emotional experiences associated with the diagnosis of cancer, the impact on the life of the patient and family as well as effects of treatment	97%	100%
Knowledge on health belief theories and health behaviour change theories.	78%	76%
Family planning and fertility implications	NEW in 2	90%
Psycho-social support	NEW in 2	88%
Ability to identify ethical, ethnic/ancestral, cultural, religious, legal, fiscal, and societal issues related to understanding health and genetic information	77%	86%
Demonstrate use of a range of effective communication skills/strategies to provide information, psychological and emotional support to individuals and communities about cancer	96%	96%
Select and adopt an appropriate communication approach, from a range of core communication and consultation skills, to effectively support the people with high risk of cancer	95%	96%
Ability to communicate and support family members at risk	NEW in 2	92%
Nurses' role in the follow up/support	NEW in 2	92%

There were also 16 comments with recommendations for future education. As there were many topics for training, experts recommended dividing the training into three themes (genetics, behaviour, and communication) or into different levels of training (essential, intermediate, and advanced training). Others commented on the delivery methods (proposing case studies and online learning). The results and a summary of the comments were sent to the participants.

4. Discussion

This study served to identify the competencies that cancer nurses should obtain in cancer genetics and cancer prevention. The main competencies were defined by a Delphi panel of international experts in cancer and genetics. This is the first time that a study aimed to develop a consensus on the competencies needed in cancer, genetics and prevention, and communication barriers. The multidisciplinary panel rapidly reached a high level of consensus, with all participants recognising the importance of developing oncology nurses' competencies in these fields.

There have been studies focused on creating competencies in cancer and genomics for healthcare professionals, but most were not specific to nurses and did not incorporate individual risk reduction behaviours and health promotion skills. A recent study developed competencies of healthcare professionals (including nurses) in cancer genomics [35]. This study focused on knowledge, attitudes, and skills needed for nurses and physicians, identifying 42 items just in cancer and genomics. We looked at the items described in that study and found that all necessary items were reflected in the competencies of the present study. In that study, the only item for prevention was the "Ability to use health promotion/disease prevention practices that incorporate genetic and genomic information as well as personal and environmental risk factors" in brackets; however, the authors focused more on preventive management than personal behaviours, adding "giving advice and discussing preventive management such as mammography, colonoscopy". Their Delphi panel was also limited to a small sample of six geneticists and doctors, failing to include nurses in a study that aimed to develop nursing competencies [35]. Cancer nurses require education tailored to their needs and competencies, especially when it comes to their role in following up with these individuals after diagnosis [20,26].

People with a genetic predisposition to cancer see different healthcare professionals but generally have little engagement in health promotion interventions, as their knowledge of cancer predisposition syndromes is limited [36]. There is a need to develop more interventions for these patients and measure multiple outcomes, including health behaviours and changes as well as the person's wellbeing. Cancer prevention and education to improve health literacy is important for the population but even more so for those who are already worried about their cancer risk [37]. To make this possible, healthcare professionals have to improve their own knowledge on cancer genetics, prevention strategies, and behaviours that affect cancer risk, but they also need to understand and support behavioural change [38]. Recent studies show how these patients generally mistrust the information they receive from healthcare professionals about their genetic syndrome and about prevention and screening practices, as they are not informed of what a cancer predisposition to a pathogenic variant means [36,39].

Cancer nurses are well positioned to evaluate and support these individuals and their families, and there is a need for them to assume this role to favour the mainstream implementation of genetics in oncology. A multiprofessional study examining the activities of nurses in cancer screening identified patient education and health promotion as core activities of these nurses [40]. Interestingly, physicians could envision the potential for nurses' activities in research, patient education and evaluation of interventions more clearly than nurses themselves. The author speculated that social desirability bias could have affected this result, as physicians were reporting what nurses could provide while the nurses were describing what they were actually doing in practice.

The competencies obtained in the present study lay the foundation for the development of cancer genetics and cancer prevention training for cancer nurses. The expert feedback highlighted the importance of cancer nurses developing their knowledge, knowing the role of genetic counsellors, working together, and the involvement of experts in patient follow-up. Genetic counsellors have and will continue to play a key role in helping patients and families before and after testing as well as supporting oncology healthcare professionals in mainstreaming this model. These professionals play a crucial part in educating healthcare professionals in genetics and have unique knowledge on counselling.

Strengths and Limitations

The main limitations of the study are related to the Delphi method. Despite being an effective tool to reach consensus and to plan educational curricula, this technique is subject to bias [29,30,35]. In this case, the competencies proposed in the survey were based on a previous systematic review. While this is very helpful, it may also limit participants' discussion and ideas, affecting the validity by influencing the first part of the survey [31,41]. For this reason, the authors did leave room for participants to add their suggestions and encouraged their ideas and participation, which improved the second round. Moreover, the anonymity of the Delphi technique removed any group pressure, reducing the potential for spurious changes in answers [33]. Another common limitation of Delphi studies [41], which also applies to this one, is the attrition rate. Even though participants received reminder emails, and the second round was left open longer than initially planned, the attrition rate between the first and second round was 31%.

However, the study method also offers strengths. The Delphi technique is useful in that it allows for the involvement of different expert professionals from various countries. As this study is part of the exploratory phase of a larger project, this engagement enriches the project with multiprofessional and multicountry expertise [31,33]. The Delphi process and anonymity was followed rigorously. This study was also piloted before the implementation of the first round, something highly recommended for Delphi studies but not commonly followed [41].

Another strength resides in the composition of the expert panel. Apart from the relatively large sample size in both rounds, panellists derived from numerous countries (especially in Europe), and the results were strengthened by their expertise. Methodological guidance suggests Delphi studies seek similar representation from each area of expertise, and this study the panel was very evenly divided into the following three backgrounds: healthcare professionals working in genetics, healthcare professionals looking after cancer patients, and researchers in cancer and genetics [27]. It also had representation from different healthcare professionals. The high rate of agreement and similar consensus rates obtained in the two rounds suggest stability, making the results more reliable [27].

While the expert panel offered many strengths, the composition of professionals, with 82.2% from European countries, poses a limitation as the results may not represent what educational needs other countries from outside Europe may have on these topics. Though professionals from outside of Europe participated on this consensus study, experts from outside of Europe could be consulted to ensure the validity of the educational programme outside the represented countries. Future studies should also look at patients' views on the education required for cancer nurses.

5. Conclusions

The introduction of genomics in oncology has changed all aspects of cancer care and provides many prevention opportunities. Healthcare professionals have demonstrated interest in involving cancer nurses, raising their awareness of the importance of genetics, and training them to incorporate cancer prevention in their daily practice.

This Delphi study aimed to achieve consensus on the competencies needed for a cancer nurse education programme in cancer genetics and cancer prevention. The results of this study, which involved experts from all around the world, show that healthcare professionals

consider it important for cancer nurses to increase their knowledge and skills on these topics. This study will serve as the basis for the content of the educational programme to promote cancer and genetics and cancer prevention education for cancer nurses.

Author Contributions: Conceptualization and methodology, C.D.d.l.R.d.l.S., P.F.-O. and T.L-C.; the Delphi and pilot and first analysis with anonymisation of the data, C.D.d.l.R.d.l.S.; analysis and thematic synthesis C.D.d.l.R.d.l.S., P.F.-O. and T.L.-C.; writing—original draft preparation and graphics, C.D.d.l.R.d.l.S.; writing—review and editing, P.F.-O.; supervision, P.F.-O. and T.L.-C. All authors have read and agreed to the published version of the manuscript.

Funding: This research received no external funding.

Institutional Review Board Statement: The study was conducted in accordance with the Declaration of Helsinki and approved by the Ethics Committee of University of Barcelona (protocol code IRB00003099) on 8 July 2021.

Informed Consent Statement: Informed consent was obtained from all subjects involved in the study.

Data Availability Statement: Not applicable.

Acknowledgments: Acknowledge to those that help us to pilot and gave feedback to improve the Delphi, and Meggan for the professional proofread.

Conflicts of Interest: The authors declare no conflict of interest.

References

1. Merlo, L.M.F.; Pepper, J.W.; Reid, B.J.; Maley, C.C. Cancer as an Evolutionary and Ecological Process. *Nat. Rev. Cancer* **2006**, *6*, 924–935. [CrossRef] [PubMed]
2. Nagy, R.; Sweet, K.; Eng, C. Highly Penetrant Hereditary Cancer Syndromes. *Oncogene* **2004**, *23*, 6445–6470. [CrossRef] [PubMed]
3. Buchanan, A.H.; Lester Kirchner, H.; Schwartz, M.L.B.; Kelly, M.A.; Schmidlen, T.; Jones, L.K.; Hallquist, M.L.G.; Rocha, H.; Betts, M.; Schwiter, R.; et al. Clinical Outcomes of a Genomic Screening Program for Actionable Genetic Conditions. *Genet. Med.* **2020**, *22*, 1874–1882. [CrossRef] [PubMed]
4. Laforest, F.; Mann, B.; Edwards, A.; Kirkegaard, P. Genetic Cancer Risk Assessment in General Practice: Systematic Review of Tools Available, Clinician Attitudes, and Patient Outcomes. *Br. J. Gen. Pract.* **2019**, *69*, E97–E105. [CrossRef]
5. Grzymski, J.J.; Elhanan, G.; Morales Rosado, J.A.; Smith, E.; Schlauch, K.A.; Read, R.; Rowan, C.; Slotnick, N.; Dabe, S.; Metcalf, W.J.; et al. Population Genetic Screening Efficiently Identifies Carriers of Autosomal Dominant Diseases. *Nat. Med.* **2020**, *26*, 1235–1239. [CrossRef]
6. Maxwell, K.N.; Domchek, S.M. Cancer Treatment According to BRCA1 and BRCA2 Mutations. *Nat. Rev. Clin. Oncol.* **2012**, *9*, 520–528. [CrossRef]
7. Cragun, D.; Beckstead, J.; Farmer, M.; Hooker, G.; Dean, M.; Matloff, E.; Reid, S.; Tezak, A.; Weidner, A.; Whisenant, J.G.; et al. IMProving Care After Inherited Cancer Testing (IMPACT) Study: Protocol of a Randomized Trial Evaluating the Efficacy of Two Interventions Designed to Improve Cancer Risk Management and Family Communication of Genetic Test Results. *BMC Cancer* **2021**, *21*, 1–11. [CrossRef]
8. Madlensky, L.; Trepanier, A.M.; Cragun, D.; Lerner, B.; Shannon, K.M.; Zierhut, H. A Rapid Systematic Review of Outcomes Studies in Genetic Counseling. *J. Genet. Couns.* **2017**, *26*, 361–378. [CrossRef]
9. Riley, B.D.; Culver, J.O.; Skrzynia, C.; Senter, L.A.; Peters, J.A.; Costalas, J.W.; Callif-Daley, F.; Grumet, S.C.; Hunt, K.S.; Nagy, R.S.; et al. Essential Elements of Genetic Cancer Risk Assessment, Counseling, and Testing: Updated Recommendations of the National Society of Genetic Counselors. *J. Genet. Couns.* **2012**, *21*, 151–161. [CrossRef]
10. Eggert, J. Genetics and Genomics in Oncology Nursing: What Does Every Nurse Need to Know? *Nurs. Clin.* **2017**, *52*, 1–25. [CrossRef]
11. Schneider, J.L.; Goddard, K.A.B.; Muessig, K.R.; Davis, J.V.; Rope, A.F.; Hunter, J.E.; Peterson, S.K.; Acheson, L.S.; Syngal, S.; Wiesner, G.L.; et al. Patient and Provider Perspectives on Adherence to and Care Coordination of Lynch Syndrome Surveillance Recommendations: Findings from Qualitative Interviews. *Hered. Cancer Clin. Pract.* **2018**, *16*, 1–10. [CrossRef]
12. De Simone, L.M.; Arjunan, A.; Vogel Postula, K.J.; Maga, T.; Bucheit, L.A. Genetic Counselors' Perspectives on Population-Based Screening for BRCA-Related Hereditary Breast and Ovarian Cancer and Lynch Syndrome. *J. Genet. Couns.* **2021**, *30*, 158–169. [CrossRef] [PubMed]
13. Singer, C.F.; Balmaña, J.; Bürki, N.; Delaloge, S.; Filieri, M.E.; Gerdes, A.M.; Grindedal, E.M.; Han, S.; Johansson, O.; Kaufman, B.; et al. Genetic Counselling and Testing of Susceptibility Genes for Therapeutic Decision-Making in Breast Cancer—an European Consensus Statement and Expert Recommendations. *Eur. J. Cancer* **2019**, *106*, 54–60. [CrossRef] [PubMed]

14. Collins, I.M.; Steel, E.; Mann, G.B.; Emery, J.D.; Bickerstaffe, A.; Trainer, A.; Butow, P.; Pirotta, M.; Antoniou, A.C.; Cuzick, J.; et al. Assessing and Managing Breast Cancer Risk: Clinicians' Current Practice and Future Needs. *Breast* **2014**, *23*, 644–650. [CrossRef] [PubMed]
15. Keogh, L.A.; Steel, E.; Weideman, P.; Butow, P.; Collins, I.M.; Emery, J.D.; Mann, G.B.; Bickerstaffe, A.; Trainer, A.H.; Hopper, L.J.; et al. Consumer and Clinician Perspectives on Personalising Breast Cancer Prevention Information. *Breast* **2019**, *43*, 39–47. [CrossRef]
16. Williams, J.K.; Katapodi, M.C.; Starkweather, A.; Badzek, L.; Cashion, A.K.; Coleman, B.; Fu, M.R.; Lyon, D.; Weaver, M.T.; Hickey, K.T. Advanced Nursing Practice and Research Contributions to Precision Medicine. *Nurs. Outlook* **2016**, *64*, 117–123. [CrossRef]
17. Seven, M.; Pasalak, S.I.; Guvenc, G.; Kok, G. Knowledge Level and Educational Needs of Turkish Oncology Nurses Regarding the Genetics of Hereditary Breast and Ovarian Cancer. *J. Contin. Educ. Nurs.* **2017**, *48*, 570–576. [CrossRef]
18. Prolla, C.M.D.; Da Silva, P.S.; Oliveira Netto, C.B.; Goldim, J.R.; Ashton-Prolla, P. Knowledge about Breast Cancer and Hereditary Breast Cancer among Nurses in a Public Hospital. *Rev. Lat. Am. Enferm.* **2015**, *23*, 90–97. [CrossRef]
19. Matsumoto, M.; Sasaki, N.; Tsukigawa, Y.; Otsubo, R.; Yano, H.; Nagayasu, T. A Survey of the Awareness and Educational Needs of Nurses in Nagasaki Prefecture Regarding Hereditary Breast and Ovarian Cancer. *J. Cancer Educ.* **2022**, 1–6. [CrossRef] [PubMed]
20. Jacobs, C.; Rahman, B. One Size Does Not Fit All: The Case for Targeted Education in Genetics and Genomics for Cancer Nurses. *Eur. J. Cancer Care* **2021**, *30*, e13480. [CrossRef]
21. Hickey, K.T.; Taylor, J.Y.; Barr, T.L.; Hauser, N.R.; Jia, H.; Riga, T.C.; Katapodi, M. Nursing Genetics and Genomics: The International Society of Nurses in Genetics (ISONG) Survey. *Nurse Educ. Today* **2018**, *63*, 12–17. [CrossRef]
22. Skirton, H.; Barnoy, S.; Erdem, Y.; Ingvoldstad, C.; Pestoff, R.; Teksen, F.; Williams, J. Suggested Components of the Curriculum for Nurses and Midwives to Enable Them to Develop Essential Knowledge and Skills in Genetics. *J. Community Genet.* **2012**, *3*, 323–329. [CrossRef] [PubMed]
23. Zureigat, B.; Gould, D.; Seven, M. Educational Interventions to Improve Nurses' Competency in Genetics and Genomics: A Scoping Review. *J. Contin. Educ. Nurs.* **2022**, *53*, 13–20. [CrossRef] [PubMed]
24. Charalambous, A.; Wells, M.; Campbell, P.; Torrens, C.; Östlund, U.; Oldenmenger, W.; Kelly, D. A scoping review of trials of interventions led or delivered by cancer nurses. *Int. J. Nurs. Stud.* **2018**, *86*, 36–43. [CrossRef]
25. The EONS Cancer Nursing Education Framework. Available online: https://cancernurse.eu/education/cancer-nursing-education-framework/ (accessed on 10 June 2022).
26. Rahman, B.; McEwen, A.; Phillips, J.L.; Tucker, K.; Goldstein, D.; Jacobs, C. Genetic and Genomic Learning Needs of Oncologists and Oncology Nurses in the Era of Precision Medicine: A Scoping Review. *Per. Med.* **2022**, *19*, 139–153. [CrossRef] [PubMed]
27. Drumm, S.; Bradley, C.; Moriarty, F. 'More of an Art than a Science'? The Development, Design and Mechanics of the Delphi Technique. *Res. Soc. Adm. Pharm.* **2022**, *18*, 2230–2236. [CrossRef] [PubMed]
28. Avella, J.R. Delphi Panels: Research Design, Procedures, Advantages, and Challenges. *Int. J. Dr. Stud.* **2016**, *11*, 305–321. [CrossRef]
29. Broomfield, D.; Humphris, G.M. Using the Delphi Technique to Identify the Cancer Education Requirements of General Practitioners. *Med. Educ.* **2001**, *35*, 928–937. [CrossRef]
30. Kirk, M.; Tonkin, E.; Skirton, H. An Iterative Consensus-Building Approach to Revising a Genetics/Genomics Competency Framework for Nurse Education in the UK. *J. Adv. Nurs.* **2014**, *70*, 405–420. [CrossRef]
31. Niederberger, M.; Spranger, J. Delphi Technique in Health Sciences: A Map. *Front. Public Health* **2020**, *8*, 457. [CrossRef]
32. Diez de los Rios de la Serna, C.; Fernandez-Ortega, P.; Lluch-Canut, T. CN13 Review of Healthcare Interventions to Promote Cancer Prevention by Improving Lifestyle Behaviours. *Ann. Oncol.* **2021**, *32*, S1260. [CrossRef]
33. Shariff, N.J. Utilizing the Delphi Survey Approach: A Review. *J. Nurs. Care* **2015**, *4*, 246. [CrossRef]
34. Okoli, C.; Pawlowski, S.D. The Delphi Method as a Research Tool: An Example, Design Considerations and Applications. *Inf. Manag.* **2004**, *42*, 15–29. [CrossRef]
35. Hoxhaj, I.; Tognetto, A.; Acampora, A.; Stojanovic, J.; Boccia, S. Core Competencies in Cancer Genomics for Healthcare Professionals: Results From a Systematic Literature Review and a Delphi Process. *J. Cancer Educ.* **2021**, 1–11, Epub ahead of print. [CrossRef] [PubMed]
36. Warner, N.Z.; Gleeson, C.; Fahey, P.; Horgan, R.; Groarke, A.M. Experiences of Living with Lynch Syndrome: A Reflexive Thematic Analysis. *Eur. J. Oncol. Nurs.* **2022**, *58*, 102117. [CrossRef]
37. Mazor, K.M.; Roblin, D.W.; Williams, A.E.; Greene, S.M.; Gaglio, B.; Field, T.S.; Costanza, M.E.; Han, P.K.J.; Saccoccio, L.; Calvi, J.; et al. Health Literacy and Cancer Prevention: Two New Instruments to Assess Comprehension. *Patient Educ. Couns.* **2012**, *88*, 54. [CrossRef]
38. Mahon, S.M. Providing Care for Previvors: Implications for Oncology Nurses. *Clin. J. Oncol. Nurs.* **2014**, *18*, 21–24. [CrossRef]
39. Campbell-Salome, G.; Buchanan, A.H.; Hallquist, M.L.G.; Rahm, A.K.; Rocha, H.; Sturm, A.C. Uncertainty Management for Individuals with Lynch Syndrome: Identifying and Responding to Healthcare Barriers. *Patient Educ. Couns.* **2021**, *104*, 403–412. [CrossRef]

40. Benito, L.; Binefa, G.; Vidal, C.; Lluch, M.T.; Puig, M.; Padrol, I.; García, M. Identifying Nursing Activities in Population-Based Colorectal and Breast Cancer Screening Programs in Spain: A Delphi Study. *Collegian* **2017**, *24*, 351–359. [CrossRef]
41. Keeney, S.; Hasson, F.; McKenna, H.P. A Critical Review of the Delphi Technique as a Research Methodology for Nursing. *Int. J. Nurs. Stud.* **2001**, *38*, 195–200. [CrossRef]

Article 3:

Title	Hereditary cancer syndrome carriers: feeling left in the corner
Authors	Diez de los Rios de la Serna, C., Fernández-Ortega, P., Lluch-Canut, T
Reference	Study accepted. Letter of Acceptance in Annex
Journal	Seminars in Oncology nursing
Keywords	Hereditary Cancer Syndromes, Cancer Nurses, Hereditary Breast and Ovarian Cancer, Lynch Syndrome, Patient Experiences, Hereditary Cancer
Impact Factor	2.2
Category	Oncology (nursing)
Quartile	Q2
ISSN	07492081, 18783449

Seminars in Oncology Nursing 000 (2024) 151624

Contents lists available at ScienceDirect

Seminars in Oncology Nursing

journal homepage: https://www.journals.elsevier.com/seminars-in-oncology-nursing

Hereditary Cancer Syndrome Carriers: Feeling Left in the Corner

Celia Diez de los Rios de la Serna[a,*], Maria Teresa Lluch-Canut[a], Maria Paz Fernández-Ortega[a,b]

[a] PhD Programme, Faculty of Nursing, Bellvitge Campus, University of Barcelona (UB), Barcelona, Spain
[b] Institut Català d'Oncologia (ICO) Barcelona, Barcelona, Spain

ARTICLE INFO

Key Words:
Hereditary cancer syndromes
Cancer nurses
Hereditary breast and ovarian cancer
Lynch syndrome
Patient experiences
Hereditary cancer

ABSTRACT

Objectives: There is limited evidence on health promotion interventions in people with hereditary cancer syndromes or on their main sources of support and information. This study aimed to understand these patients' experiences and needs, including their information needs, their views on prevention and mental health, and the support they want from nurses.
Methods: This qualitative study included 22 people (8 previvors and 14 survivors) with hereditary breast and ovarian syndrome or Lynch syndrome from 10 European countries. Participants underwent individual semi-structured interviews, which were recorded and transcribed for reflexive thematic analysis. The patient and public involvement panel provided input on study design and thematic analysis.
Results: Patient experiences were similar regardless of the country and access to testing and screening. Participants reported receiving little information on the importance of health behaviors for cancer risk and expressed their wish to be followed by cancer professionals. They felt compelled to seek support and information from the internet and patient groups. The main themes identified were: (unmet) informational and support needs, seeing life in a different way, and limitations of health care providers.
Conclusions: People with hereditary cancer syndromes need professionals to be involved in their long-term management and to provide reliable information. As genomics are increasingly integrated in oncology, the need for professionals to support these populations will increase.
Implications for Nursing Practice: Nurses are crucial for promoting self-management and advocating for patient decision-making; however, they need skills and knowledge to do so. There is a need for nurses to get more involved in understanding hereditary cancer syndromes and an opportunity to take the lead in the care of these people.

People with hereditary cancer syndromes (HCSs) have a high risk of developing certain cancers in their lifetime due to a pathogenic gene mutation that confers an increased susceptibility to cancer.[1] The most common HCSs are hereditary nonpolyposis colorectal cancer, also called Lynch syndrome, which entails a higher risk of mainly colon, endometrial, gastric, and small intestine cancer; followed by hereditary breast and ovarian cancer (HBOC), which primarily increases the risk of breast, ovarian and prostate cancer.[2] Even though HCSs are not common, they are linked to around 10% of cancers.[3] Diagnosis is essential in order to plan for long-term follow-up, as HCSs affect entire families, entailing a 50% chance of passing on mutated genes to offspring and a high risk of early onset cancer.[2]

Genetic testing is gradually being implemented in clinical practice, as early diagnosis of these syndromes allows for preventive controls to decrease risk and detect cancer early.[4,5] Interventions include behavioral counselling, surveillance, prophylactic medication, surgeries, and possible modifications in the management and treatment of any cancer that develops.[6] People with HCSs thus require comprehensive, personalized care for them and their families, which poses challenges for the health care system and the person affected alike. HCS carriers have to understand their diagnosis and its implications, get recommendations about preventive controls, be prepared for complex and personal decisions (eg, regarding preventive surgery), and usually take responsibility for disclosing the results to the rest of their family.[7,8] Numerous studies have looked at the psychological impact of HCS diagnosis and related decision-making, which is associated with uncertainty, doubts, and distress.[9-13] These patients have often reported a lack of follow-up and poorly integrated care; they describe being attended by health care professionals with little knowledge of HCSs, leaving them feeling lonely, with nobody to turn to.[10]

The implications of communications with family and the feelings of guilt associated with this process have also been explored.[10,14] People

* Address correspondence to: Celia Diez de los Rios de la Serna, MSc, RN, ANP, Faculty of Nursing University of Barcelona Carrer de la Feixa Llarga, s/n, 08907 L'Hospitalet de Llobregat, Barcelona. Spain
E-mail addresses: cdiezdde7@alumnes.ub.edu, celiadiezrios@gmail.com (C. Diez de los Rios de la Serna).

https://doi.org/10.1016/j.soncn.2024.151624

Layperson Summary

What we investigated and why

There are very little studies looking at how actions to improve lifestyle affect people with genetic alterations that increase their risk of having cancer. There is also very little information on where they find information and support. The aim of this study was to understand the needs of people with these genetic alterations and how nurses can help.

How we did our research

We did interviews with 22 people with these genetic alterations (8 never had cancer and 14 had cancer in the past) from 10 different European country. We asked them to have an interview that were recorded and analyzed. Some people with these genetic alterations helped to plan the study, the questions and how we could analyze them.

What we have found

People have similar experiences and problems no matter where they came from. They want more information on lifestyles and they also want to have cancer professionals to follow them up. They also told us they look for information in internet and patients' groups. Our main findings were: they want to be understood, they see life in a different way and they want more from health care professionals.

What it means

After looking at the evidence is important to suggest having professionals that know about genetic alterations to answer their questions and worries. Nurses are probably the best professionals to do it but they need to know more about it as this need is only going to grow.

with an HCS are urged to notify at-risk relatives, so it is left to those with the syndrome to disclose the risks and the implications to family members, often without the guidance of health care professionals.[6]

From the health system perspective, guidelines for the standard of care in HCS carriers, for example, those of the National Comprehensive Cancer Network (NCCN) or the European Society of Medical Oncology, give advice on testing criteria, risk reduction strategies (including screening and surveillance), and reproductive risks.[15-18] However, they rarely address the personal needs or actions of people affected by HCSs, such as psychosocial support or lifestyle behaviors. Guidelines for genetic counselling do acknowledge the need for multidisciplinary team work to cover the personal and social needs and provide resources to HCS carriers.[6,19] The increasing role of genetics in the cancer management poses a challenge to professionals, who may lack knowledge, skills, or confidence when providing care for people with HCSs.[20,21] Interest in educating health care professionals is growing, but this training rarely encompasses comprehensive care.

The delivery of comprehensive care can directly affect self-management and decision-making, including adherence to recommendations, management of appointments, and decision-making about risk reduction actions.[22,23] Self-management is directly affected by the person's perception of their own risk and the extent to which they believe risk reduction actions will work. Adherence to risk management recommendations is therefore not only dependent on the information that health care professionals may provide at HCS diagnosis, but the support and comprehensive care that addresses their risk perception, personal health beliefs, and the psychological effects these have.[24-26] Different studies have explored lifestyle behaviors, behavioral modification interventions,[27,28] risk perception, and psychological impacts in HCS carriers,[8,10-12] but the relationship between mental health and self-management has not been explored and is not part of the recommendations and education currently offered in oncogenetics.

The aim of this study was to explore the experience of HCS carriers (both those who have not had cancer, also known as previvors, and those who have had a cancer diagnosis) and their priorities and unmet needs regarding self-management and behavioral counselling during follow-up. A patient and public involvement (PPI) panel collaborated during the project to ensure that the study design was worthwhile for HCS carriers.[29]

Methods

This qualitative study was based on one-on-one virtual interviews with people affected by HCSs in Europe. Semi-structured interviews allow the interviewer to guide the participant to areas of interest but ensure that the flow of the interview is directed by the participant.[30] The consolidated criteria for reporting qualitative research[31] were used to report this study (Appendix 1).

Recruitment

Recruitment was done through posts on social media inviting eligible participants to contact the principal investigator (PI). Posts were promoted in patient support groups and by PPI panel members and were shared by carriers and hereditary cancer professionals via Twitter, Facebook, and LinkedIn. Inclusion criteria were: diagnosis of an HCS (Lynch syndrome or HBOC), regardless of whether any cancer had been diagnosed, residence in a European country, and being able to speak Spanish or English. People who were undergoing cancer treatment at the time of the study, or who were not confirmed carriers of an HCS, were excluded. We used purposive sampling to recruit a demographically and geographically heterogeneous European sample,[32] as data saturation was not considered the appropriate way to determine the sample size.[33] Instead, the final sample was determined by the quality of data.

Interested individuals were sent information about the study, a participant information sheet, and a consent form. Those who replied that they were willing to be part of the study were contacted to set a date for their interviews. The PI conducted the interviews using the university Zoom account. The investigators did not know any of the participants prior to the study. All participants understood the aim of the study, gave written informed consent to take part in the interviews, and agreed to be recorded during the interviews (audio and video). They were informed that only partial quotes from the interviews were going to be used under a pseudonym, and only for study purposes. The university ethics committee approved the study (IRB00003099).

Data Collection

Interviews were conducted from January to June 2022 and lasted 40 to 70 minutes. Audio recordings were transcribed by the PI, using Sonix.ai to facilitate transcription. Field notes, comments, and observations were added to the transcription by the PI.

The interview questions were open and covered questions about different domains: living with high risk of cancer and understanding how participants perceived and managed their risk in terms of lifestyle, mental health, and communication with health care professionals (Table 1). The interview guide was underpinned by a variety of theoretical perspectives informed by nursing and psychology theory. The research team considered the role of mental health in self-care to be important, along with self-care tools in patients at risk, especially in oncology. We also considered that nurses are well positioned to

TABLE 1
Interview Guide with Main Themes

Theme	Topics
Diagnosis: first contact	• When and how were you given information? Who gave it, and what information was given?
Living at risk of cancer	• Needs of yours that haven't been met since your Lynch/BRCA syndrome diagnosis • Issues prioritized in your life
Information	• What information are you missing? • Where do you get and look for the information? • What kind of information you are looking for?
Prevention	• What aspects of your life do you consider important in the risk of cancer? • What things you would like to do to prevent or decrease your risk of cancer?
Mental health	• Has there been any change in your thoughts and mental well-being as a result of the diagnosis? What emotions does it arouse in you? • How important do you think the mental/psychosocial part of the information/diagnosis and follow-up is?
Educational program	• How do you think health professionals could better meet the needs of a person with a hereditary cancer syndrome? • What do you think health professionals should know and prioritize to meet the needs of a person with a hereditary cancer syndrome?

make an impact by promoting mental health, in turn improving self-management. Orem[26] emphasizes the importance of self-care in patients at risk of cancer, Lluch[25] focuses on emotional tools as facilitators of self-care, and the Health Belief Model[24] considers the importance of personal beliefs and knowledge in their decision-making. The interview guide was developed and discussed with expert patients and was piloted by two people from the PPI panel.

Data Analysis

Two researchers independently performed reflexive thematic content analysis on the verbatim transcript, following the six-phase process.[34,35] We chose this method to identify patterns in the data and to involve research participants in the analysis. Firstly, the three authors (a nurse with experience in cancer genetic counselling and working as a research assistant, an experienced researcher working in a cancer hospital, and a professor in nursing and mental health) discussed the patterns found after independent analysis of the interviews, acknowledging their experience and bias when interpreting the transcripts.[34] The team discussed and defined the emerging themes based on the analysis and codes. This method of analysis allows the data to lead the formulation of themes in a collaborative and reflexive way that enriches the interpretation.[34,35] The themes and quotes selected were shared back with some of the participants, refined, and finalized as presented here.

All participants were offered the opportunity to review the themes developed following analysis and three participants agreed. Some expressed concern that they could be recognized based on the ID and the country, so participants' characteristics do not include the participant ID.

Results

The final sample comprised 22 individuals from 10 European countries: Denmark (n = 1, 5%), Germany (n = 3, 14%), Ireland (n = 4, 18%), the Netherlands (n = 1, 5%), Portugal (n = 2, 9%), Poland (n = 1, 5%), Slovenia (n = 2, 9%), Spain (n = 3, 14%), Turkey (n = 1, 5%), and the United Kingdom (n = 4, 18%). There were 14 people (64%) affected by HBOC (10 previously diagnosed with cancer and 4 previvors), and 8 (36%) affected by Lynch syndrome (5 previously diagnosed with cancer and 2 previvors). Seven participants were men (32%), 19 (86%) had a family history of cancer, and 15 (68%) had a personal history of cancer. Among those diagnosed with cancer, four (27%) knew about the HCS prior to developing a tumor. Participants were diagnosed with their HCSs between 2000 and 2021. Their characteristics are displayed in Table 2.

Three main themes were developed: (1) (unmet) informational and support needs, (2) seeing life in a different way, and (3)

TABLE 2
Participant Characteristics

Code	Sex	Family history	Personal history	Discovered pre- or postcancer	Cancer	Year DX mutation	Preventive measures	Follow-ups	Behavioral changes
B1	F	Yes	Yes	Post	Breast	2021	Tamoxifen, Mast, BSO	BT, USS	No
L1	M	Yes	Yes	Pre	Bowel	2010	Aspirin	OGD, COL, dermatologist	Yes
B2	M	Yes	Yes	Post	Breast, prostate	2011	Tamoxifen	PSA	No
L2	F	No	Yes	Post	Bowel, breast, pancreas, melanoma, endometrium	2020	Aspirin, tamoxifen, BSO	COL	No
B3	F	Yes	No	N/A	N/A	2019	No	MRI	No
L3	F	Yes	Yes	Post	Endometrial, bowel, breast	1996	No	COL	No
B4	F	Yes	Yes	Post	Ovary	2015	No	MMG, MRI	No
L4	F	Yes	No	N/A	N/A	2017	Partial excision of colon	COL, OGD dermatologist	Yes
B5	F	Yes	No	N/A	N/A	2008	Mast, TAH, BSO	No	Yes
L5	F	Yes	Yes	Post	Bowel	2011	No	COL	Yes
B6	F	Yes	Yes	Post	Breast	2012	Double mast	MRI, USS, BT	Yes
L6	M	Yes	No	N/A	N/A	2019	No	COL	No
B7	F	Yes	Yes	Post	Breast	2013	No	MRI and USS	Yes
L7	M	No	Yes	Post	Bowel	2012	No	COL	No
B8	F	Yes	Yes	Post	Ovary	2012	No	MRI and USS	No
L8	M	Yes	No	N/A	N/A	2018	No	COL	Yes
B9	M	Yes	No	N/A	N/A	2019	Mast, BSO	No	No
B10	F	Yes	No	N/A	N/A	2011	No	Yes	No
B11	M	No	Yes	Pre	Breast	2014	No	PSA	Yes
B12	F	Yes	Yes	Pre	Breast	2019	No	MRI and USS	Yes
B13	F	Yes	Yes	Pre	Breast	2013	Mast	Bex, USS	No
B14	F	Yes	Yes	Post	Breast	2000	Mast, TAH, BSO	No	No

Bex, breast examination; BSO, bilateral salpingo-oophorectomy; BT, blood test; COL, colonoscopy; Mast, mastectomy; MMG, mammography; MRI, magnetic resonance imaging; N/A, not applicable; OGD, gastroscopy; PSA, prostate-specific antigen; TAH, total abdominal hysterectomy; USS, ultrasound.

limitations of health care providers. Each theme is further divided into various subthemes (Fig.), as described below with quotes (L identifies a person with Lynch syndrome and B identifies a person with HBOC).

Theme 1: Unmet Informational and Support Needs

This theme represents the experiences of HCS carriers related to accessing information and support from health care professionals or others. Respondents perceived deficiencies in follow-up care and felt frustration around the need for psychosocial support and information about the implications of the risks and risk reduction techniques. Participants described how they went about looking for information (usually on the internet) and their feeling of relief upon finding others with similar experiences.

Subtheme 1: Finding the "Right" Information

Participants felt that while the information they received during the diagnosis of their HCS was good and generally well supported by a geneticist or genetic counsellor, their real information needs started after they had known/accepted the results. This meant that when they had questions they did not know where to go.

> I've got to say, whilst the counselling was very good, I don't think I totally took it in and realised the implications. Then the questions started but no one was there anymore. **B2**

People living with Lynch syndrome generally felt more distress, as they found a complete lack of information outside the field of genetics.

> [talking about if they got information] No, not a thing. I went to the library. I couldn't find anything.**L3**

Subtheme 2: A Cancer Patient, But Not a Cancer Patient

There was a difference in follow-up between those who had cancer prior to a diagnosis of HCS and those who had never had cancer, as the latter felt like they had the risks, follow-up, and fear but could not access the same services cancer patients could.

> I have a follow-up with my oncologist every six months, but that's not because I've got Lynch, that's almost like a by-product . . . I think I am very lucky to have all of these professionals that follow me up. **L2 (Personal history of cancer)**

> I'm not what's classified as a healthy person. I'm classified as a cancer patient, and I will be for the rest of my life, but I cannot access specific care for cancer because I haven't had an active invasive cancer yet**. L4 (previvor)**

Subtheme 3: Understanding the Dimensions of Risk

Regardless of their syndrome or cancer status, participants all agreed that they did not receive any information on their risks of developing cancers other than the ones primarily affected by their HCSs. Many were not even sure what risks of other cancers they had or if there was any follow-up for these.

> I didn't even realise at the time that I've got an increased chance of getting prostate cancer. **B2**

> The problem I see with Lynch is that it can affect all different parts of your body, so there's no one person to go to. I don't have a Lynch consultant to control all my cancer risks. **L6**

Those who had risk-reducing surgeries perceived a lack of subsequent follow-up and support. A couple of participants explained their fear of not having anyone to go to as they had no further follow-ups, and another participant commented on the risks of other cancers.

> I feel like nobody addresses any of the other cancers. It's like, oh, no, it's not going to happen to you. Don't waste our time with this. And you are kind of left like, well, what do I do? What do I look out for? And they're like, No, you're fine. You've got this surgery done and you have nothing else to worry about. **B9**

They also felt as if health care professionals guided them to the follow-ups and risk-reducing surgeries without fully explaining all the implications.

> They [healthcare professionals] need to prepare the patient for the consequences. Not only what this mutation brings. But also, what are the consequences if you remove your breasts? If you remove your ovaries? If you remove your tubes? How hard are the surgeries? I knew nothing about that. **B13**

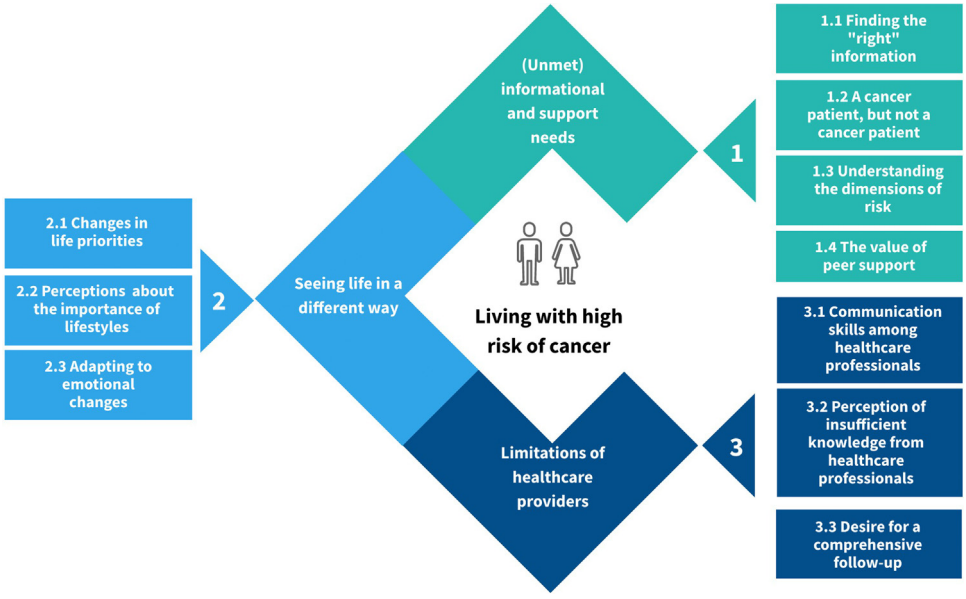

FIG. Themes and subthemes.

There is another part they do not inform you, the effects of the surgery on you. **B1**

Subtheme 4: The Value of Peer Support

Participants found support and understanding in patient support groups. This made them feel more confident about the reliability of the information they were reading, as they felt that knowledgeable people were responsible for curating it. But more than anything, they went to groups to feel understood and to find others with similar worries.

They have a huge amount of support, so I learn a lot from there. **L6**

So, I thought, I have to talk to somebody, how they deal with what are they doing, what they are thinking about, how are they dealing with the risk, knowing that they could be sick many times and again and again, that was really hard for me. **B14**

There was a difference between people who took an active role in patient support groups and those who were more passive and felt they were just consumers of information. Those in active roles were more critical of where the information they were seeking came from and felt they could go to other health care professionals (as they built connections via social media) to resolve their doubts. They also felt very helpful when giving others the support that they had trouble finding themselves.

I found a lot of comfort going into the role of like peer support . . . to give what I did not get. **B9**

At the same time, participants with a more active role in patient supports group felt the responsibility and challenge of supporting others and at times felt they could not share their own burdens and concerns.

When I go to Facebook groups and in my family, I'm the support, they assume I know what I am talking about. I am not afraid of it (BRCA), but I can't ask them questions, as I am the one that needs to know. **B8**

Theme 2: Seeing Life in a Different Way

The lives of people with HCSs change in a personal and emotional way. Participants had to adjust to the diagnosis, faced decisions on risk reduction, and pondered whether they were healthy and how to be healthier. Throughout the interviews, they also discussed the emotional challenges of living with risk, both for themselves and for their family members.

Subtheme 1: Changes in Life Priorities

Many participants mentioned changes in their perspective about life after being diagnosed. For previvors, this meant a new adjustment to life, with some feeling it did not change their life for the better or give them a more positive perspective and others experiencing it as an opportunity to value life more.

I do see things differently, probably especially in the last, probably the last year. **L6**

I try and have more family time, enjoy life a bit more and stress less. I guess it is similar to what would happen with any other major impact on your health. **B12**

For survivors, the diagnosis did not involve the same adjustment, as they felt that their cancer diagnosis was more important than the HCS diagnosis in terms of re-evaluating their life priorities.

Not really, nothing changed, I guess the shock was bigger when I got the cancer diagnosis (. . .) but still I don't think I made any big changes apart from what I could not do. **L2**

Subtheme 2: Perceptions About the Importance of Lifestyles

When asking about prevention, participants recounted that they had thought about health behaviors, but generally they did not assign much importance to it.

For the most part, participants felt they had received little information about lifestyle and did not see it necessary to make any changes, or they thought that they had such a high risk that behavioral modifications would not impact their cancer risk.

The doctor didn't tell me anything about lifestyles . . . I didn't change my lifestyle. **B2**

The doctor said, smoking is not good, you know, but it doesn't cause breast cancer. And, why change? I will probably have cancer at some point. **B14**

In contrast, some participants felt like the HCS status was a motivation to either maintain good habits or improve on them, even if just to face a future cancer from a level of good baseline health.

I changed my diet, my life, and after the surgery [preventive mastectomy] I started to go to the swimming pool twice a week. **B6**

I think it's important to be in good health to face cancer. **L1**

Subtheme 3: Adapting to Emotional Changes

Emotions change and evolve over time in those with HCSs.

I do feel like a different person. From how I was. And, yeah, I think, a change sort of emotionally. On the one hand, I feel quite resilient. And on the other hand, I feel quite vulnerable. **L6**

Participants were very attuned to the changes, but they perceived very little support toward their psychosocial wellbeing. One patient even expressed her gratitude to the interviewer.

I have never been asked about my feelings, thank you. **B5**

Their coping strategies also varied depending on how long ago they were diagnosed with their HCS, whether they had developed cancer, and the idea they had about HCSs. Most participants moved from the shock of diagnosis:

It was all a lot to take on board. **B11**

. . . to a state of uncertainty they described as a *double-edged sword, a loaded die,* or a *bad lottery,* which made them live with fear.

I cannot have back pain; it has to be cancer. **B4**

Some expressed feelings of defeat after so much constant surveillance.

Year after year, the same fight to get an appointment, the preparation for a colonoscopy, which is horrible . . . sometimes I just want to stop. **L3**

Some participants felt relief upon diagnosis of HCSs, something that they almost expected. This experience was normally associated with a long family history of cancer.

In some way, for me, it was it was quite a relief. **L2**

I was now glad that "I now know my enemy". **L7**

Others felt that HCS was a constant burden, and they needed support.

> I think they're both equally hard, really living, getting the diagnosis wasn't nice, and then living with it. **B10**

> I basically I feel like no one was there to guide me and to hold my hand. **L1**

Theme 3: Limitations of Health Care Providers

This theme encompasses perceived shortcomings in health care professionals' knowledge and communication skills around HCSs (as opposed to unmet informational support needs reported in theme 1). They described difficulties when facing different professionals and a desire for a less disjointed health care experience.

Subtheme 1: Communication Skills Among Health Care Professionals

Most participants described communication with health care professionals about HCSs as a real, unmet need, with many wishing for more active listening and an empathetic attitude from their health care providers.

> You have to listen, what is this person thinking about now? What are they understanding now? What is the knowledge now? What do they know? What do they not know? And then listen. **B14**

> They're great at telling us how often we need a colonoscopy and how often we need to have guidance screening. But they're not really good at sitting and listening and explaining what you want to know. **L4**

Subtheme 2: Perception of Insufficient Knowledge from Health Care Professionals

Participants felt that health professionals lacked an understanding of HCSs. There was a common feeling of deficits in communication in the health care system, which created a burden for patients, as they had to repeatedly share information about their condition to different professionals.

> Any time I spoke to somebody medically and said I had Lynch syndrome, they looked so lost themselves. I think they thought I was making it up. So, I stopped telling anybody. **L4**

> I think it's terrible, though, that it comes down to the luck of the draw. Whether you are lucky enough to meet a medical professional that knows enough about it. **L8**

While they understood that health care professionals cannot know everything, they wished their providers knew more about HCSs. This need was more relevant for people with Lynch syndrome, who found that many health care professionals had not heard about this syndrome, while HBOC carriers felt that the BRCA mutation was understood, but only in terms of breast and ovarian risks, not other cancer risks. They found it difficult to secure the follow-up appointments recommended for their syndrome, meaning that the patients themselves had to be on top of their own surveillance, as there is no coordination within the health care system.

> You know, you have to fight for some things. I mean, fight to get a colonoscopy every two years, even though that is standard. Because some doctors will be like,... Oh, well, you know, you don't need it now. **L5**

> *I have my gynaecologist. Yeah. So, uh, he's not, he's not interested either. So it's totally up to me to make sure that I'm tested regularly.* ***B3***

> *I also was not given any sort of plan, monitoring plan or whatever it was.* **L1**

Subtheme 3: Desire for a Comprehensive Follow-Up

Participants—especially HBOC previvors and those with Lynch syndrome—commonly wished they had a health professional who would be available at their follow-ups. On many occasions, they identified nurses and specifically nurses working in oncology as their ideal person to talk to on a regular basis and use as a focal point with the health care system in case any doubts arose.

> People feel lost. We feel that there is a need to have an annual check-up with a consultant or nurse. Some of us are scared, even though we know there's a very low chance of finding anything after mastectomy. **B5**

Interviewees wished that health providers would give them a list of services, support groups, and written information to help them navigate their needs and queries. Another participant recommended health professionals use social media to reach young people.

> For example, a health professional could tell you the news and say, we've got this website with this information, and this is what you can do to reduce your risks, and be very clear and supportive. **L6**

> You can and give guidance and counselling and education, getting involved in social media like an internal app. **B7**

Discussion

Our results indicate that people with HCSs from around Europe perceive that health professionals do not have complete and exhaustive knowledge about HCSs and, consequently are not providing the support they need. While the information needs of previvors and those that have had cancer are different,[9] they all reported unmet needs. They received some information pre- and post-test but little to no information thereafter, regardless of the country where they were receiving care. Other studies, both in and outside of Europe, have drawn similar conclusions about the need for information.[10,36]

On top of their unmet needs in the information domain, they also described lack of coordination in their follow-up, difficulties that have also been shared in other studies.[11,37] HCS carriers feel unsupported, and this affects their ability to cope with the diagnosis and their decision-making. During the interviews, they sometimes wondered if their decisions would have been different had they received more information.

Participants also expressed an interest in having cancer nurses involved in their care. These nurses generally perform a lot of educational interventions, but the programs are normally targeted to people undergoing cancer treatment, not to previvors, and the education mainly focuses on the side effects of treatment in survivors.[38] HCS carriers differ from the general patient population, who generally demonstrate great trust in their health care professionals, especially nurses,[39] in that their trust in health care professionals is undermined by their perceived lack of knowledge about HCSs.[9]

In line with these perceived shortfalls, HCS carriers seek information and peer support elsewhere, especially the internet, where they often find solace from peers affected by the same syndrome. Our study also suggests that knowledge shared by peers with similar experiences is greatly valued. Health professionals should recognize that value providing information that people need and facilitating access to support groups. Social media platforms have become popular avenues to seek health care information and support among

cancer patients.[40] There is a lack of research on how HCS support groups and social media are used by this population and the impact it has on them. In 2016, a couple of genetic counsellors commented on the benefits of social media and support groups[41] and the need for health care professionals to get involved in social media,[42] but there has been no subsequent research on interventions or impact. A Cochrane Review in breast cancer patients and support group interactions[43] concluded that being part of a support group can relieve anxiety and even improve quality of life, and in interviews with HBOC previvors,[44] participants shared that writing down their experience was helpful to process their own feelings. In this study, we found that HCS carriers felt a sense of belonging when participating in these groups, but there was also some sense of burnout in those providing the most support. While many studies have explored the benefits, few have investigated the negative impact of social media; one qualitative study in young cancer patients[45] revealed that they felt some level of burden and negative impact from reading the experience of others. Future research should explore the role of social media for HCS carriers as well as the emotional burden of supporting peers.

Participants had very different views on the perceived benefits of their own actions such as lifestyle behaviors. Health care professionals have the ability to influence those beliefs,[24] but while health professionals are knowledgeable about cancer prevention, they do not promote literacy on cancer prevention and lifestyle behaviors.[46,47] The current lack of engagement from health care professionals in follow-up and health behaviors, together with the dearth of behavioral research and interventions to address lifestyle behaviors,[27,28] is affecting the self-management and actions of HCS carriers. There is also a lost opportunity regarding the potential to use social media and patient support groups to promote cancer prevention and healthy behaviors.[48,49]

In order to feel engaged in their self-management and self-care, HCS carriers need to have their psychosocial needs met and be able to accept and process the storm of feelings brought on by an HCS diagnosis. Participants in the interviews, and the existing evidence, reinforce the need to improve patients' experience.[9-12,36,50] Regardless of whether they have been diagnosed with cancer, finding out about a genetic alteration of this kind takes some getting used to.[37,51] From assimilating the concerns from and about their family to understanding the myriad impacts of the different management strategies they are offered, HCSs carriers have a real need for psychosocial support,[51] a trusting relationship with the health care system, and health services that promote healthy behaviors. In this line, the six factors formulated in the Multifactorial Positive Mental Health Model[25]: personal satisfaction, prosocial attitude, self-control, autonomy, problem-solving and self-actualizations, and interpersonal relationships; have been proven effective in different intervention programs,[52,53] with a positive impact on self-care.[26]

Our findings are suggestive of a generalized need for more nurse education on HCSs in Europe. Health care professionals should be more involved in the follow-up of HCS carriers, who in turn need to be empowered to take a lead role in their own care. Closer involvement of health systems in satisfying these needs would allow patients to feel more supported and empowered.

Study Strengths and Limitations

A key strength of this study was the involvement of a PPI panel during the planning and design stage. PPI is very important in cancer research and more so in PhD projects to ensure that the studies and research questions are pertinent for them.[28,54,55] Our inclusion of both previvors and survivors also means our results are generalizable to all HCS carriers, without neglecting the differences that may exist according to their cancer status or country. While systems, opportunities, and access to genetic counselling vary in these countries,[56] HCS carriers have common needs and experiences.

This study also has some limitations. Participants were recruited via social media and patient support groups, so the views and needs of HCS carriers that do not even have the information and support from these groups are not included; therefore, we may be leaving out an important group to explore. Also, while we had the views of both men and women with BRCA and Lynch syndrome, far fewer men were in our sample, meaning we may have overlooked some of their needs by not including a large enough sample.

Conclusions and Clinical Implications for Nurses

This qualitative study provides insight into the perspectives and needs of HCS carriers on their long-term management. People with HCSs need a health professional they can go to in order to ask questions and who can help them navigate the system and meet their needs. Moreover, health care professionals should have a role in the follow-up and long-term management of these patients. Nurses are well placed to promote self-management and advocate for patient decision-making; however, they need to have adequate skills and knowledge to effectively perform this role.

In light of how many of our participants were actively supporting others on social media, future studies should look further into the involvement of HCS carriers in social media and the emotional burden that they feel.

People with HCSs are asked to make very difficult decisions on surveillance and management. Building professional capacity and conducting more research on lifestyle behaviors and behavioral theories would help enable these patients to make informed choices. With the rapid adoption of genomics in cancer care, there is and will be more demand for genetic testing, which will increase the need for professionals to guide and support this population.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

CRediT authorship contribution statement

Celia Diez de los Rios de la Serna: Conceptualization, Methodology, Validation, Formal analysis, Investigation, Resources, Data curation, Writing – original draft, Writing – review & editing, Visualization, Project administration, Funding acquisition. **Maria Teresa Lluch-Canut:** Conceptualization, Methodology, Validation, Resources, Writing – original draft, Writing – review & editing, Supervision. **Maria Paz Fernández-Ortega:** Conceptualization, Methodology, Validation, Formal analysis, Resources, Data curation, Writing – original draft, Writing – review & editing, Supervision.

Funding

This work was supported by an education grant award from the International Society of Nurses in Genetics (ISONG).

Acknowledgments

The authors would like to sincerely thank the PPI panel for their support, ideas and encouragement. The authors are also immensely grateful to the participants who were so open and shared their experiences and struggles. The authors also want to thank all the professionals, patients and patient organizations that supported and shared this study.

Supplementary materials

Supplementary material associated with this article can be found in the online version at doi:10.1016/j.soncn.2024.151624.

References

1. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med*. 2015;17(5):405–424. https://doi.org/10.1038/GIM.2015.30.
2. Imyanitov EN, Kuligina ES, Sokolenko AP, et al. Hereditary cancer syndromes. *World J Clin Oncol*. 2023;14(2):40. https://doi.org/10.5306/WJCO.V14.I2.40.
3. Garutti M, Foffano L, Mazzeo R, et al. Hereditary cancer syndromes: a comprehensive review with a visual tool. *Genes (Basel)*. 2023;14(5):1025. https://doi.org/10.3390/GENES14051025/S1.
4. De Keersmacker S, Meder S, Cassidy D. Europe's beating cancer plan: a new EU approach to prevention, treatment and care. *Press Release Eur Comm*. 2021:(February):1–3.
5. Berger MF, Mardis ER. The emerging clinical relevance of genomics in cancer medicine. *Nat Rev Clin Oncol*. 2018;15(6):353–365. https://doi.org/10.1038/s41571-018-0002-6.
6. Riley BD, Culver JO, Skrzynia C, et al. Essential elements of genetic cancer risk assessment, counseling, and testing: updated recommendations of the National Society of Genetic Counselors. *J Genet Couns*. 2012;21(2):151–161. https://doi.org/10.1007/s10897-011-9462-x.
7. Knapp ST, Revette A, Underhill-Blazey M, et al. MyLynch: a patient-facing clinical decision support tool for genetically-guided personalized medicine in Lynch syndrome. *Cancers (Basel)*. 2023;15(2):391. https://doi.org/10.3390/CANCERS15020391/S1.
8. Dean M, Fisher CL. Uncertainty and previvors' cancer risk management: understanding the decision-making process. *J Appl Commun Res*. 2019;47(4):460–483. https://doi.org/10.1080/00909882.2019.1657236.
9. Dean M, Scherr CL, Clements M, Koruo R, Martinez J, Ross A. "When information is not enough": a model for understanding BRCA-positive previvors' information needs regarding hereditary breast and ovarian cancer risk. *Patient Educ Couns*. 2017;100(9):1738–1743. https://doi.org/10.1016/J.PEC.2017.03.013.
10. Warner NZ, Gleeson C, Fahey P, Horgan R, Groarke AM. Experiences of living with Lynch syndrome: a reflexive thematic analysis. *Eur J Oncol Nurs*. 2022;58: 102117. https://doi.org/10.1016/J.EJON.2022.102117.
11. Warner NZ, Groarke AM. A qualitative reflexive thematic analysis into the experiences of being identified with a BRCA1/2 gene alteration: "So many little, little traumas could have been avoided.". *BMC Health Serv Res*. 2022;22(1):1–12. https://doi.org/10.1186/S12913-022-08372-W/TABLES/2.
12. Bartley N, Napier C, Best M, Butow P. Patient experience of uncertainty in cancer genomics: a systematic review. *Genet Med*. 2020;22(9):1450–1460. https://doi.org/10.1038/S41436-020-0829-Y.
13. Oliveira FFB, de Barros Silva PG, de Sant'Ana RO, et al. How does genetic testing influence anxiety, depression, and quality of life? A hereditary breast and ovarian cancer syndrome suspects trial. *Support Care Cancer*. 2020;29(7):3521–3530. https://doi.org/10.1007/S00520-020-05867-2.
14. Pollard S, Kalloger S, Weymann D, et al. Genetic testing for hereditary cancer syndromes: patient recommendations for improved risk communication. *Health Expect*. 2020;23(4):884–892. https://doi.org/10.1111/hex.13062.
15. Stjepanovic N, Moreira L, Carneiro F, et al. Hereditary gastrointestinal cancers: ESMO Clinical Practice Guidelines for diagnosis, treatment and follow-up. *Ann Oncol*. 2019;30(10):1558–1571. https://doi.org/10.1093/annonc/mdz233.
16. National Comprehensive Cancer Network. NCCN genetic/familial high-risk assessment: breast, ovarian, and pancreatic. Available at: https://www.nccn.org/professionals/physician_gls/pdf/genetics_bop.pdf Accessed February 23, 2024.
17. National Comprehensive Cancer Network. NCCN genetic/familial high-risk assessment: colorectal. Available at: https://www.nccn.org/professionals/physician_gls/pdf/genetics_colon.pdf Accessed February 23, 2024.
18. Balmaña J, Balaguer F, Cervantes A, Arnold D. Familial risk-colorectal cancer: ESMO clinical practice guidelines. *Ann Oncol*. 2013;24(Suppl.6):vi73–vi80. https://doi.org/10.1093/annonc/mdt209.
19. Berliner JL, Cummings SA, Boldt Burnett B, Ricker CN. Risk assessment and genetic counseling for hereditary breast and ovarian cancer syndromes—practice resource of the National Society of Genetic Counselors. *J Genet Couns*. 2021;30(2):342–360. https://doi.org/10.1002/JGC4.1374.
20. Calzone KA, Kirk M, Tonkin E, Badzek L, Benjamin C, Middleton A. Increasing nursing capacity in genomics: overview of existing global genomics resources. *Nurse Educ Today*. 2018;69:53. https://doi.org/10.1016/J.NEDT.2018.06.032.
21. Papaioannou K, Kampourakis K. Health-care professionals' awareness and understanding of genomics. *Appl Genomics Public Heal*. 2020:225–242. https://doi.org/10.1016/B978-0-12-813695-9.00012-1.
22. Connors LM, Voian N, Shi Y, Lally RM, Edge S. Decision making after BRCA genetic testing. *Clin J Oncol Nurs*. 2014;18(3):E58–E63. https://doi.org/10.1188/14.CJON.E58-E63.
23. Moreno L, Linossi C, Esteban I, et al. Germline BRCA testing is moving from cancer risk assessment to a predictive biomarker for targeting cancer therapeutics. *Clin Transl Oncol*. 2016;18(10):981–987. https://doi.org/10.1007/s12094-015-1470-0.
24. Glanz K, Rimer BK, Viswanath K, eds. *Health Behavior and Health Education: Theory, Research, and Practice*. 4th ed. San Francisco: Jossey-Bass; 2008.
25. Lluch Canut MT. *Construcción de una escala para evaluar la salud mental positiva. Doctoral thesis*. Barcelona, Spain: Faculty of Psychology, University of Barcelona; 1999.
26. Naranjo Hernández Y, Concepción Pacheco JA, Rodríguez Larreynaga M. The Self-Care Deficit Theory: Dorothea Elizabeth Orem. *Gac Méd Espirit*. 2017;19(3):89–100.
27. Donato KM, Moore K, Parker WM, et al. Health and lifestyle behaviors in colorectal cancer survivors with and without Lynch syndrome. *J Community Genet*. 2019;11(1):59–63. https://doi.org/10.1007/s12687-019-00421-y.
28. Diez de los Rios de la Serna C, Fernández-Ortega P, Lluch-Canut T. Lifestyle behavior interventions for preventing cancer in adults with inherited cancer syndromes: systematic review. *Int J Environ Res Public Health*. 2022;19(21). https://doi.org/10.3390/ijerph192114098.
29. Briefing notes for researchers - public involvement in NHS, health and social care research | NIHR. Available at: https://www.nihr.ac.uk/documents/briefing-notes-for-researchers-public-involvement-in-nhs-health-and-social-care-research/27371 Accessed July 27, 2023.
30. Kallio H, Pietilä AM, Johnson M, Kangasniemi M. Systematic methodological review: developing a framework for a qualitative semi-structured interview guide. *J Adv Nurs*. 2016;72(12):2954–2965. https://doi.org/10.1111/JAN.13031.
31. Tong A, Sainsbury P, Craig J. Consolidated criteria for reporting qualitative research (COREQ): a 32-item checklist for interviews and focus groups. *Int J Qual Health Care*. 2007;19(6):349–357. https://doi.org/10.1093/INTQHC/MZM042.
32. Robinson OC. Sampling in interview-based qualitative research: a theoretical and practical guide. *Qual Res Psychol*. 2014;11(1):25–41. https://doi.org/10.1080/14780887.2013.801543.
33. Braun V, Clarke V. To saturate or not to saturate? Questioning data saturation as a useful concept for thematic analysis and sample-size rationales. *Qual Res Sport Exerc Heal*. 2021;13(2):201–216. https://doi.org/10.1080/2159676X.2019.1704846.
34. Braun V, Clarke V. One size fits all? What counts as quality practice in (reflexive) thematic analysis? *Qual Res Psychol*. 2020;18(3):328–352. https://doi.org/10.1080/14780887.2020.1769238.
35. Braun V, Clarke V, Hayfield N, Terry G. Thematic analysis. *In: Handbook of Research Methods in Health Social Sciences*. Singapore: Springer; 2019:843–860. https://doi.org/10.1007/978-981-10-5251-4_103.
36. Wellman ML, Holton AE, Kaphingst KA. "Where do I go? Who do I go to?": BRCA previvors, genetic counselors and family planning. *PEC Innov*. 2023;2:100157. https://doi.org/10.1016/J.PECINN.2023.100157.
37. Campbell-Salome G, Buchanan AH, Hallquist MLG, Rahm AK, Rocha H, Sturm AC. Uncertainty management for individuals with Lynch syndrome: identifying and responding to healthcare barriers. *Patient Educ Couns*. 2021;104(2):403–412. https://doi.org/10.1016/J.PEC.2020.07.017.
38. Charalambous A, Wells M, Campbell P, et al. A scoping review of trials of interventions led or delivered by cancer nurses. *Int J Nurs Stud*. 2018;86:36–43. https://doi.org/10.1016/J.IJNURSTU.2018.05.014.
39. Dinç L, Gastmans C. Trust in nurse–patient relationships. *Nurs Ethics*. 2013;20(5):501–516. https://doi.org/10.1177/0969733012468463.
40. Gandamihardja TAK, Liyanage S, Coutee T, Peled AW, Masannat YA. The role of social media and breast cancer: how does it impact patients? *Breast Care*. 2023;18(3):203–208. https://doi.org/10.1159/000530433.
41. Stefansdottir V. Experience of social media support group for BRCA carriers. *J Genet Couns*. 2016;25(6):1342–1344. https://doi.org/10.1007/S10897-016-0009-Z.
42. Gallagher L, McCuaig J, Benoit L, Davies C. It's time for the genetic counseling profession to embrace social media. *J Genet Couns*. 2016;25(6):1338–1341. https://doi.org/10.1007/S10897-016-9950-0/METRICS.
43. Mccaughan E, Parahoo K, Hueter I, Northouse L, Bradbury I. Online support groups for women with breast cancer. *Cochrane Database Syst Rev*. 2017;2017(3):CD011652. https://doi.org/10.1002/14651858.CD011652.pub2.
44. Wellman ML, Holton AE, Kaphingst KA. Previvorship posting: why breast cancer previvors share their stories on social media. *Health Commun*. 2023;38(11):2441–2449. https://doi.org/10.1080/10410236.2022.2074780.
45. Stage C, Klastrup L, Hvidtfeldt K. Ugly media feelings: Negative affect in young cancer patients' experiences of social media. *First Monday*. 2021;26(7). https://doi.org/10.5210/fm.v26i7.11093.
46. Fleary SA, Paasche-Orlow MK, Joseph P, Freund KM. The relationship between health literacy, cancer prevention beliefs, and cancer prevention behaviors. *J Cancer Educ*. 2019;34(5):958–965. https://doi.org/10.1007/S13187-018-1400-2.
47. Albada A, Vernooij M, van Osch L, Pijpe A, van Dulmen S, Ausems MGEM. Does and should breast cancer genetic counselling include lifestyle advice? *Fam Cancer*. 2014;13(1):35–44. https://doi.org/10.1007/S10689-013-9672-5.
48. Lauckner C, Whitten P. The differential effects of social media sites for promoting cancer risk reduction. *J Cancer Educ*. 2016;31(3):449–452. https://doi.org/10.1007/s13187-015-0881-5.
49. Han CJ, Lee YJ, Demiris G. Interventions using social media for cancer prevention and management: a systematic review. *Cancer Nurs*. 2018;41(6):E19–E31. https://doi.org/10.1097/NCC.0000000000000534.
50. Dean M, Davidson LG. Previvors' uncertainty management strategies for hereditary breast and ovarian cancer. *Health Commun*. 2018;33(2):122–130. https://doi.org/10.1080/10410236.2016.1250187.
51. Yanes T, Willis AM, Meiser B, Tucker KM, Best M. Psychosocial and behavioral outcomes of genomic testing in cancer: a systematic review. *Eur J Hum Genet*. 2019;27(1):28–35. https://doi.org/10.1038/s41431-018-0257-5.
52. Puig Llobet M, Sánchez Ortega M, Lluch-Canut M, Moreno-Arroyo M, Hidalgo Blanco MÀ, Roldán-Merino J. Positive mental health and self-care in patients with chronic physical health problems: implications for evidence-based practice. *Worldviews Evidence-Based Nurs*. 2020;17(4):293–300. https://doi.org/10.1111/WVN.12453.

53. Sánchez-Ortega MA, Lluch-Canut MT, Roldán-Merino J, et al. Nursing intervention to improve positive mental health and self-care skills in people with chronic physical health conditions. *Int J Environ Res Public Health*. 2022;20(1):528. https://doi.org/10.3390/IJERPH20010528.
54. Colomer-Lahiguera S, Steimer M, Ellis U, et al. Patient and public involvement in cancer research: a scoping review. *Cancer Med*. 2023;12(14):15530–15543. https://doi.org/10.1002/cam4.6200.
55. Tanay MA, Diez de los Rios de la Serna C, Boland V, et al. Patient and public involvement in research: reflections and experiences of doctoral cancer nurse researchers in Europe. *Eur J Oncol Nurs*. 2023;64:102351. https://doi.org/10.1016/J.EJON.2023.102351.
56. Abacan MA, Alsubaie L, Barlow-Stewart K, et al. The global state of the genetic counseling profession. *Eur J Hum Genet*. 2019;27(2):183–197. https://doi.org/10.1038/s41431-018-0252-x.

Discussion

The results of each study are discussed in their respective articles. This section summarises and discusses the main findings through a broader analysis and reflection, and presents the limitations and implications for practice.

The main objective of this doctoral thesis was to investigate the needs of people living with hereditary cancer syndromes and define the educational needs of cancer nurses to support them. The constituent studies demonstrated the lack of involvement of healthcare professionals in promoting healthy lifestyles in people with HCSs; identified a number of unmet needs that HCS carriers have to deal with; brought into relief patients' desire to have comprehensive care, potentially provided by cancer nurses; and highlighted the consensus around the need for cancer nurses to have access to education in genetics, communication, and health prevention.

The involvement of the healthcare system in supporting people with HCSs could have positive impacts on the person, their health, and health services. This supportive approach has been proven effective in people with long-term conditions and in situations of self-management[160]. Regardless of whether they have a personal history of cancer, HCS carriers have surveillance and health needs and risks that may be similar to people with long-term conditions. Healthcare professionals have a role in secondary prevention for these populations.

According to nursing and psychology theories and existing evidence, nurses are the key healthcare professional to address patients' symptom management, psychological needs, and self-care requirements. This role has been extensively observed and documented in cancer patients[77,132,137,156]. Cancer nurses are the main information provider for cancer patients and therefore one of the most trusted professionals[161,162]. However, the same is not happening in HCSs. These syndromes are not currently being followed up by a particular healthcare group or specialised unit. Instead, care for this population normally focuses on surveillance and is led by different professionals depending on the cancer site. This lack of support and education leaves patients unequipped, with the sole responsibility for making health decisions and managing their risks.

One possible explanation for not having specialist follow-up care could be related to the lack of knowledge and accessibility of cancer nurses in Europe. In cancer care, there is considerable variability in access to cancer nursing education and recognition of these roles[163]. Therefore, access to specialist nurses around Europe remains quite uneven, and while some countries have better access to specialist nurses and even nurse practitioners looking after people with cancer, others do not have the same resources[80,164,165]. This has a direct impact on cancer patients, affecting their care, symptoms control, self-care abilities and wellbeing, but access to the right care can also impact survival[78]. This is also

seen in HCS care, where the involvement of cancer nurses is still nascent in a few countries, but completely absent in most others[74].

These patients report a lack of follow-up and poorly integrated care, as they do not usually have follow-up appointments with professionals knowledgeable on HCSs[75,166]. The delivery of comprehensive care can directly affect self-management and decision-making, including their beliefs, preventive actions, and attendance to follow-up[66]. Delayed diagnosis is one of the most important factors that affect patient survival and is directly impacted by the infrastructure, including nursing care and the health literacy of cancer patients[165]. But the individual's attitudes can also affect their risk, as happens with lifestyle behaviours.

The first objective related to identifying the interventions used to promote healthy lifestyles in people with high risk of cancer and assessing their effects, and this was addressed through a systematic review[167]. Since 2010, only four published articles were identified that met the inclusion criteria. Just one was focused on previvors[168], while the rest included a mix of survivors and previvors, with a predominance of survivors. This made it difficult to draw any conclusions about the need for differentiated interventions in these two groups. While their motivations might be very different, mainly due to their lived experiences, the results of the review did not allow for conclusions.

Although few interventions focus on lifestyle behaviours in HCS carriers, many studies describe modifiable behavioural risks that could be addressed in this group[94,97,99]. However, the studies from the review focused on diet, physical activity, and weight. Although tobacco is one of the modifiable lifestyle behaviours with stronger evidence linked to many cancer risks, none of the included studies addressed tobacco[5,6].

Moreover, the effects of the behavioural interventions were not the primary outcome of the included studies. The intervention effects were sometimes measured only once, and in the cases where follow-up measurements were made, the effect seemed to decline over time towards baseline levels or nearly baseline levels, calling into question the sustainability of the behavioural changes.

Only one of the included studies[168] tested interventions designed with a theoretical grounding in behavioural modification, and none measured motivation for change in terms of perceived benefit or risk perception; however, some did assess perceived barriers. There are existing models defining how to plan and implement interventions based on behaviour change theories, starting with known techniques and models[116,169]. While the theories should be taken into account to plan interventions, there is also evidence that using a theory-driven intervention does not always directly impact the behaviour intervention outcomes[170]. Sometimes this lack of effectiveness can be attributed to intervention design errors and the use of inappropriate theories[171]. Moreover, while theories can guide professionals in planning an intervention and

identifying the methods to address the related concepts, true change must be promoted by regular, appropriate health education.

An individual's health literacy greatly affects the perceived benefit of an intervention, which in turn affects their behaviour modification. Likewise, the information provided by healthcare professionals has a direct impact on patients' health literacy, with subsequent effects on their beliefs and protective attitudes[172]. People with low health literacy may be more fatalistic and therefore less inclined to believe that their actions can effectively reduce their risk[129]. A systematic review found it unlikely that genetic testing would cause a negative impact on health behaviour[169], but without appropriate recommendations, healthcare professionals are missing valuable opportunities for teachable moments to explain how important lifestyle behaviours might be, as the knowledge of risk per se does not drive behavioural changes[106].

Our review lays bare the lack of follow-up in HCS carriers and the failure to address modifiable risks, underlining the need for interventions to promote healthy lifestyle behaviours in people with HCS and provide education for both patients and nurses. In light of these findings, one could hypothesise that even if nurses were undertaking the follow-up of HCS carriers, they cannot really help the person or accompany them in all their needs without adequate training. HCS carriers need interventions that focus on health promotion, enhanced health literacy, and behavioural counselling. A comprehensive follow-up can directly mitigate the psychological blow of being diagnosed with an HCS. Theory-driven interventions performed by cancer nurses can promote more positive mental health and self-care behaviours, while helping to address the health belief of the person, with cascade effects on their lifestyle behaviours, quality of life, and self-care management.

While most experts agree that nurses have a key role in genetics, they also point out their knowledge gaps[85]. Even trained professionals with good knowledge of genetics feel they lack skills to promote behavioural change[173]. To address this gap, the second objective of this thesis was to determine what knowledge is necessary for oncology nurses to understand and be able to help people with HCS understand their cancer risk and follow a healthy lifestyle.

The systematic review showed that educational topics had to go further than genetics[60], as professionals are already missing opportunities to address lifestyle behaviours and adopt risk reduction strategies, and HCS carriers are demanding better communication and coordination of care[107]. To answer these needs, in the Delphi study we presented some suggestions to key stakeholders on topics uncovered in the systematic review around genetics and lifestyle behaviours, including existing theories, communication, and barriers. It was important to focus on competency development to enhance the capacity of the nurses to address unmet needs[174].

For the Delphi study, we recruited healthcare professionals from different backgrounds and professions with expertise in both cancer and genetics. The study had a balanced mix of healthcare professionals from different countries working in genetics, healthcare professionals looking after cancer patients, and researchers in cancer and genetics. There were also a mix of professions, including physicians, nurses, genetic counsellors, and others, who shared their views on what cancer nurses should know.

From the first round, participants agreed on the relevance of most of the proposed topics. While the three topics all started with the same number of competencies, the expert panel proposed more, especially on the topic dealing with lifestyle behaviours and their link with cancer. The level of consensus was very high – more than 80% on almost every topic/theme proposed. It is difficult to compare these results with other studies; there have been previous studies to find consensus on the knowledge needed in genetics and genomics[85] and even in cancer and genomics[175], but none linking the knowledge needs with health promotion, theory, and communication skills needed to appropriately address HCS carriers' needs.

The Delphi study focused on the perspective of healthcare providers, but it is equally important to capture HCS carriers' views and experiences. The third objective of this doctoral thesis, then, was to explore the experience of HCS carriers and their priorities and unmet needs during their diagnosis and follow-up. The qualitative study was designed to address HCS carriers' experience in general but included questions specifically addressing lifestyle behaviours, views of the role of cancer nurses, and their information needs.

Participants were people with HCSs from all around Europe, including previvors and survivors. While there were both men and women involved, the sample was mainly made up of women. Their views on the roles of cancer nurses differed somewhat, especially when comparing countries where cancer nurses have more versus less developed roles in patient care[176]. However, while the waiting times and access to genetic testing was very different from one participant to another, depending on their country, their experience of follow-up needs and sense of abandonment after diagnosis was similar. The feeling of frustration with the system when needing information and support has been identified before in HCS carriers[65]. This feeling is due to a disruption in the continuity of care, from diagnosis with the specialist genetic counsellor to negligible post-diagnostic care. The disparity does not only manifest when needing information but also in accessing the recommended follow-up, such as an annual check-up or test. Participants were happy with the information they received at diagnosis, but then they did not know where to go to find information or coordinate their follow-up. The extent of patients' unmet needs was greater for the follow-up of less prevalent cancers such as skin cancer in Lynch syndrome carriers, as their feelings of frustration were exacerbated by their provider's lack of knowledge about genetic risks[72].

When addressing lifestyle conversations and educational interventions, the results were quite negative, as very few participants reported receiving advice from their healthcare professionals about lifestyles, and those that did said this was generic, not personalised to their particular lifestyle behaviours and interests. The overall lack of engagement from healthcare professionals did not lead to consistently similar behaviours and attitudes among participants: some decided on their own to be healthier and reduce their risk of cancer; others concluded that if their healthcare professional did not mention lifestyle, it was because it was not important enough to change; and a third group considered that they would not change their lifestyle regardless of the information they received. This last group could be further subdivided: some were aware of the risks associated with their behaviours and preferred not to change it, while others felt that their genetic risk was so high that behavioural change would barely affect the overall risk of developing cancer, so it was not worth the effort. These different experiences illustrate that if healthcare professionals do not engage in conversations about health behaviours, patients have no cues for action to promote the changes[115]. Furthermore, if the healthcare professionals attending HCS carriers do not feel the need to address lifestyle[177], it is unlikely that anyone else will, which leaves this vulnerable population without vital health promotion services. Healthcare professionals' efforts in this regard (or lack thereof) may be impacted by their lack of knowledge on lifestyle risks or training in behavioural interventions.

The information needs reported by the participants went beyond lifestyles. First of all, many patients did not trust their provider's knowledge on HCSs. These patients likewise did not report trusting their cancer nurses on this topic. This finding could be attributable to many factors, including workforce issues like how many nurses are available, as well as the knowledge that nurses have on the particular problems related to the disease, treatments, and living with and HCS. However, the main reason is probably the fact that cancer nurses are not currently directly involved in care for this patient group[165].

When patients did not find answers in their health service, they looked for other sources of information and support, such as internet or patient support groups. Indeed, patients nowadays are increasingly relying on the internet and social media platforms for health information and emotional support[178]. In cancer patients, social media use is associated with unmet psychological needs, and it also represents a tool for decision-making[179]. People with HCSs also use social media to share information, personal stories, social support, and other resources. The importance of these platforms is accentuated in the case of uncommon conditions like HCSs, as patients rarely know others with the same diagnosis[180]. Nevertheless, social media also raises concerns. Trustworthiness and ethics are serious issues when accessing health information and support online, and there is a real risk that wrong or misleading information can cause more harm than good[181]. While patient support groups can be an important resource for patients, they should complement rather than replace conventional health services. The fact that cancer

patients (and HCS carriers) are not finding the necessary support from healthcare professionals is concerning and should drive changes to provide more inclusive support.

In the qualitative interviews, HCS carriers also emphasised how important it is for nurses to focus on communication and listen to them. One participant said *"if you've got cancer, then there's the specialist nurse, but not for people with hereditary syndromes. (…) I think in some ways if there was a specialist nurse perhaps to talk to, maybe I should have contacted her, I probably should have done and said, can I have a meeting with you to sort of really discuss it more what it means in person"*. These findings are also supported by the Delphi study, which showed a need and interest in cancer nurses obtaining more communication skills[182]. Compassion and communication are essential skills for cancer care[77], and people with HCS have previously highlighted unmet needs in this area – a need for more than just generic information, but rather a dialogue that takes into account their individual needs and personal journey[75]. One of the most important and clearest demands is follow-up care. While HCS carriers are not normally followed up by cancer nurses at present, nurse follow-up could address some of their communication needs. It is also important to acknowledge the growing demand for professional training and continuous education in communication for nurses[183]; this topic garnered substantial consensus in the Delphi study. Nevertheless, as discussed above with regard to professionals' needs for knowledge and skills on lifestyle and genetic risks, it is important not to presume that incorporating follow-up with cancer nurses alone would directly address this need.

Communication skills are also important for providing support when HCS carriers need to share the information with their family members[184]. In the interviews, people with family, especially those with children, felt guilt associated with possibly passing on the condition to their children and some difficulties explaining to their family members that they may also have inherited a high risk of cancer. This aspect was among the needed skills proposed in the Delphi study as well as in other studies, which have drawn attention to efforts made to improve cascade testing in family members[185]. In the interviews, many participants were hopeful about the future of their family members, as they perceived constant advances in precision medicine. They also shared the complications and guilt associated with disclosing the results. One participant said, *"it's very hard to try and tell your family about this condition, and you literally have a paragraph on a piece of paper to give to them to give their doctor".* Another lamented the lack of support in communications, saying *"It's kind of like you're given this this bomb to drop into your family and you've been given nothing at all to help you with fallout"*. These concerns and guilt associated with family members have previously been verbalised by HCSs carriers[166].

Taken together, all the studies contribute to defining the educational needs of cancer nurses. The doctoral thesis confirms a gap between what HCS carriers need and the cancer nurses' knowledge. The project also helps identify the desired competencies that

cancer nurses should have to enable the provision of comprehensive follow-up to HCS carriers.

There is a dearth of specific professional guidance for the monitoring and follow-up of HCS carriers. In 2021, a report published in Ireland showed the needs of people with HCS in that country[186]. The authors highlighted the limited access to genetic testing, which can take up to two years in Ireland. The report's findings also supported our results, describing the perception among the people affected that there is not enough support after testing and their desire for more information on risk management and communication, particularly with family members. The Irish report left the door open to addressing these needs through professionals other than genetic counsellors; this option can be generalised to outside of Ireland given the shortage of genetic counsellors worldwide[54]. The 2023 update of the ESMO guidelines on HBOC risk reduction was the first official guideline to address the unique psychological needs of people with HCSs, including their need for further information and emotional support[30]. What all the guidelines now include is the need for multiprofessional collaboration in the care of people with HCSs, including cancer nurses[29–31,34].

This doctoral thesis shows that patients want to be heard and involved in their care, but at present there are shortcomings in both theory and practice – two essential components of nursing education. While such gaps are normally related to the inadequate application of theoretical knowledge learned in university to practice[187], in the case of HCSs there are also deficiencies in theory, as genetics and genomics are not currently covered in most undergraduate and postgraduate nursing degrees[188]. At the same time, communication, health promotion, prevention, self-care, and self-management theories are part of the nursing curriculum, but this knowledge is currently not being translated into practice either. In the Delphi study, there was consensus on the need to refresh this knowledge and acquire new skills[182]. Together, the studies support conclusions on what cancer nurses should know regardless of where they work. The international sample and literature included in the reviews, from various European countries with different healthcare systems and cancer nursing roles and recognition, increase the validity of the conclusions on what is needed in Europe.

The benefits of self-care and self-management in cancer patients and in genetic counselling are known, but in order to reap those benefits, the healthcare system needs to prioritise self-care and health promotion interventions, and healthcare providers need to feel confident in delivering them[135]. The inclusion of genetics into the expertise of cancer nurses could have a direct impact on the emotional experiences of people with HCS and their families, so building their skills and competencies around addressing the needs of people with a high risk of cancer is important for the future of cancer care[59,122]. Thus, the involvement of cancer nurses in the care of people with HCS could contribute to the prevention and early diagnosis of cancer, in turn reducing the financial and social impacts of these diseases[189].

Limitations

The included studies have some methodological limitations, which have been described in each article. In addition, the project as a whole has other possible limitations and strengths, warranting some degree of caution when interpretating the results and drawing conclusions.

The studies encompassed in the thesis aim to capture and describe current needs from the perspectives of both healthcare professionals and HCS carriers. They use a cross-sectional design and include HCS carriers and healthcare professionals from across Europe. This comes with strengths but also limitations.

Including a sample from across Europe in both the Delphi study with healthcare professionals and the interviews with patients allows for comparison between what happens in different countries' healthcare systems and what patients demand, but not every country was represented. There were also some countries where HCS carriers participated in the interviews, but no healthcare professionals from that country were involved in the Delphi study, and vice versa. However, the objective of the studies was not to compare countries, but to assess the situation across Europe and explore whether HCS carriers' unmet needs were similar in different European settings.

The studies included in the systematic review were European. While country setting was not one of the inclusion criteria, this fact enables more robust conclusions that directly affect the population of study. That said, the studies did not allow for generalisation in terms of the needs of previvors, as the samples were predominated by survivors.

Study recruitment was mainly performed through social media groups and expert networks. This approach could have led to an underrepresentation of the views of people who do not feel comfortable with technology and would have been inadvertently missed. Such people could be those with fewer information resources, for example people with HCSs who do not have a patient support network, so we may be capturing only the needs of people who have more support and information, rather than those without access to these resources.

In addition, the survey and the interviews were conducted in English or Spanish, which would have left out those who may not feel comfortable in any of those languages. In the case of healthcare professionals, most of the available educational resources are in English, so we may not have been able to capture the opinion of healthcare professionals with fewer educational opportunities.

Implications for future research

The thesis identifies educational needs for cancer nurses, so the next steps should be to present the proposed educational topics and outcomes to a panel of experts in education to inform curriculum planning for cancer nurses, including the length of training and the methods of delivery and evaluation. This should be followed by piloting the educational intervention for cancer nurses.

Another interesting research question for future studies has to do with the design of a behavioural intervention targeting lifestyle behaviours, planned with experts and PPI, and taking into consideration the theoretical framework to promote healthy behaviours. The design should be followed by a pilot and work to design interventions that could be maintained and supported over time to promote long-term outcomes with reinforcement of healthy attitudes.

The qualitative studies capture the experience of HCS carriers at one time point. The use of longitudinal studies using more quantitative tools, such as patient-reported outcomes measures (PROMs), could also help to identify the unmet needs of this population and how they change over time. As one of the limitations was not including people less active on social media, it would also be of interest to try to actively engage hospitals and genetic services to support enrolment.

Some work has already been done to improve communication between healthcare professionals and patients in genomics. The doctoral candidate was invited to participate in a scoping review to identify current gaps in communicating the results of biomarker testing in tumours, which on many occasions prompts the decision to perform HCS testing. This research – done with healthcare professionals, patient advocates and patients – also identified the need for knowledgeable and confident healthcare professionals to get involved in conversations and provide information to patients even before testing[190]. That study supports the results of the doctoral thesis and the need for further professional training in communication and genomics.

Recommendations for service development

The following recommendations derive directly from the study results, the participants' views, and suggestions from the patient and public involvement panel.

The most important recommendation that emerged is to develop comprehensive follow-up opportunities to support people with HCS. This would require training professionals according to the competencies defined in the present study in order to ensure that services respond to the unmet needs of HCS carriers. This training should consider the importance of including HCS carriers as experts in the training planning and delivery.

Today, there is a strong interest in cancer and genetics, reflected in the diversity of countries represented in the included samples. Oncology societies should explore opportunities for partnerships and potential licencing arrangements with educational institutions to make training as accessible as possible.

Involvement of patients and the public in research and in the development of education programmes is challenging, particularly in multilingual contexts. When designing European training resources, the translation of development materials can help ensure that a diverse panel of stakeholders informs the development of the programme. It is also important to consider translating guidelines and training programmes to increase access to these in professionals' native languages.

While knowledge and competencies related to communication, behaviours, and health promotion are part of nursing degrees, there should be continuous education offered for nurses to continue practicing and building their expertise and familiarity with these competencies.

There is also a need to incorporate genetics, genomics, and precision medicine into nursing degrees. Until then, these topics should at least be covered through continuous training for cancer nurses.

Potential benefits to patients and the healthcare system

With the exponential increase of genomics in cancer care and expanding access to testing, it is logical to predict that an increasing number of people will be diagnosed with HCSs. Thus, healthcare systems need to be adapted to support their adequate management. Having knowledgeable and confident cancer nurses who are actively involved in the care of HCS carriers could:

- improve shared decision-making in consultations with patients;
- ensure effective support and up-to-date advice for HCS carriers and their families (whether already cancer patients or only those at risk) ;
- empower HCS carriers and increase self-management behaviours;
- improve the mental health and decrease the anxiety of people with HCSs;
- reduce the morbidity and mortality of people with HCSs by increasing early diagnosis and addressing lifestyle behaviours;
- increase family communication, allowing for early detection of healthy individuals with an HCS;
- improve the adherence to recommendations like attendance to surveillance and healthier lifestyle behaviours;
- facilitate appropriate referral to other professionals when needed;
- promote family planning and fertility advice according to the best evidence and resources available in each country;
- improve health system engagement on cancer risks in people with HCSs.

This approach could help reduce the risk of cancer and, if cancer develops, improve early detection, with the subsequent positive financial implications stemming from reductions in cancer morbidity and mortality.

Conclusions

The main conclusions of this doctoral thesis are:

1. Healthcare professionals do not regularly promote healthy lifestyles in people at high risk of cancer. The interventions identified focus on diet and exercise and are short in duration, with short-term benefits. In people with HCS, care should start by understanding the person's beliefs regarding lifestyles and include tailored health promotion, addressing all modifiable cancer risks.
2. There is a need for accessible education in genomics and continuous training in health behaviours and communication for cancer nurses around Europe.
3. People with hereditary cancer syndromes have numerous unmet needs, especially related to accessing information and support. This population should have access to the guidance and support needed after diagnosis, including the implications of genetic testing, family communication, and prevention strategies.
4. The healthcare system should create services with trained cancer nurses to coordinate comprehensive follow-up in people with hereditary cancer syndromes.

Dissemination

Each of the studies has been presented in conferences and sent for publication. A summary of the dissemination channels of the thesis is available in Table 3.

The systematic review was presented at EONS14 conference at ESMO 2021 Congress and was published in the *International Journal of Environmental Research and Public Health*.

The Delphi study was shared as a poster-presentation at the 55th conference of the European Society of Human Genetics (ESHG) and published in the *Journal of Personalized Medicine*.

The results of the qualitative study were disseminated through a poster-presentation at the 56th ESGH conference and in a presentation at the 2023 International Conference on Cancer Nursing (ICCN) and published in *Seminars in Oncology Nursing*.

The whole thesis project was also presented in a seminar at the European Oncology Nursing Society (EONS) conference EONS16 and at ESMO 2023 in a presentation called 'Health promotion, prevention and screening'.

The doctoral candidate and her supervisor, Dr Fernández-Ortega, were also invited to be part of an editorial[191] and an opinion paper[192] discussing the role of cancer nurses in cancer prevention.

Table 3. Scientific dissemination from thesis

Objective	Method	Study	Conference presentation
Objective 1: To identify which interventions are used to promote a healthy lifestyle in people at risk of cancer.	Systematic review	Lifestyle Behavior Interventions for Preventing Cancer in Adults with Inherited Cancer Syndromes: Systematic Review.[167]	Presentation at EONS14 CN13-Review of healthcare interventions to promote cancer prevention by improving lifestyle behaviours[193]
Objective 2: Determine what knowledge is necessary for oncology nurses to understand and be able to help people with HCS understand their cancer risk and improve their health-related behaviours.	Delphi Study	Educational Programme for Cancer Nurses in Genetics, Health Behaviors and Cancer Prevention: A Multidisciplinary Consensus Study[182]	Poster at 55th European Society of Human Genetics (ESHG) EP23.010 Educational Programme on Genetics, Lifestyle Behaviours and Cancer Prevention A Multidisciplinary Consensus Study[194]
Objective 3: Explore the experience of hereditary cancer syndrome carriers and their priorities and unmet needs during their diagnosis and follow-up.	Qualitative study	Accepted in Seminars in Oncology Nursing	Poster at 56th ESHG EP13.009 Experiences and needs for those with hereditary cancer syndromes: "What they say on internet or in hereditary supportive groups" Presentation at ICCN 2023 "I ask internet or patient groups" Hereditary Cancer Syndromes carriers experience
PhD project	Presented as an invited speaker at EONS16 conference at ESMO 2023		

Funding

The doctoral thesis project received an educational grant (Figure 5 with no Grant number) from the International Society of Nurses in Genetics (ISONG), which supported the transcription of the interviews and the dissemination of the results in the ICCN conference.

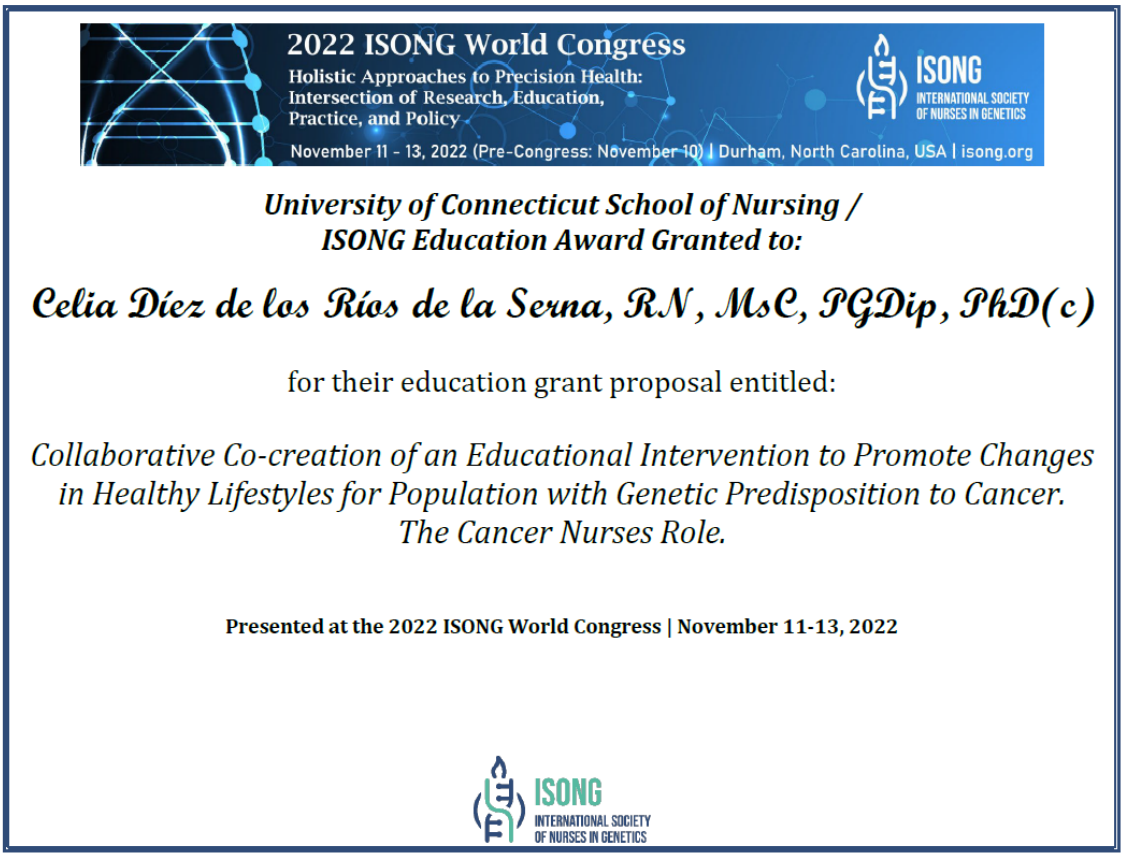

Figure 5. ISONG grant award

Also, the Colegio Oficial de Enfermeros y Enfermeras de Barcelona provided reimbursement for the open access publication of the Delphi study and the registration of the ESGH conferences.

The doctoral student also obtained support to finance the professional language review of the systematic review thanks to the Sol·licitud d'ajuts per a la traducció íntegra o revisió d'articles per la publicació en revistes indexades JCR per alumnes del programa de doctorat en infermeria I salut de la Universitat de Barcelona – Convocatòria Desembre 2021.

References

1. Sung H, Ferlay J, Siegel RL, Laversanne M, Soerjomataram I, Jemal A, et al. Global Cancer Statistics 2020: GLOBOCAN Estimates of Incidence and Mortality Worldwide for 36 Cancers in 185 Countries. *CA Cancer J Clin*. 2021;71(3):209-249. doi:10.3322/CAAC.21660

2. Ferlay J, Ervik M, Lam F, Colombet M, Mery L, Piñeros M, et al. Global Cancer Observatory: Cancer Today. 2024 Lyon, France: International Agency for Research on Cancer. Accessed January 20, 2023. https://gco.iarc.who.int/today

3. Horgan D, Baird AM, Middleton M, Mihaylova Z, Van Meerbeeck JP, Vogel-Claussen J, et al. How Can the EU Beating Cancer Plan Help in Tackling Lung Cancer, Colorectal Cancer, Breast Cancer and Melanoma? *Healthc 2022, Vol 10, Page 1618*. 2022;10(9):1618. doi:10.3390/HEALTHCARE10091618

4. Europe's Beating Cancer Plan Communication from the Commission to the European Parliament and the Council. European Commission: Brussels, Belgium, 2020. Accessed January 15, 2023. https://health.ec.europa.eu/system/files/2022-02/eu_cancer-plan_en_ 0.pdf

5. Whiteman DC, Wilson LF. The fractions of cancer attributable to modifiable factors: A global review. *Cancer Epidemiol*. 2016;44(2016):203-221. doi:10.1016/j.canep.2016.06.013

6. Soerjomataram I, Bray F. Planning for tomorrow: global cancer incidence and the role of prevention 2020–2070. *Nat Rev Clin Oncol*. 2021;18(10):663-672. doi:10.1038/s41571-021-00514-z

7. Marzo-Castillejo M, Vela-Vallespín C, Bellas-Beceiro B, Bartolomé-Moreno C, Melús-Palazón E, Vilarrubí-Estrella M, et al. Recomendaciones de prevención del cáncer. Actualización PAPPS 2018. *Aten Primaria*. 2018;50:41-65. doi:10.1016/S0212-6567(18)30362-7

8. Visscher BB, Steunenberg B, Heijmans M, Hofstede JM, Devillé W, Van Der Heide I, et al. Evidence on the effectiveness of health literacy interventions in the EU: A systematic review. *BMC Public Health*. 2018;18(1):1-12. doi:10.1186/S12889-018-6331-7/TABLES/2

9. Berger MF, Mardis ER. The emerging clinical relevance of genomics in cancer medicine. *Nat Rev Clin Oncol*. 2018;15(6):353-365. doi:10.1038/s41571-018-0002-6

10. Hanahan D. Hallmarks of Cancer: New Dimensions. *Cancer Discov*. 2022;12(1):31-46. doi:10.1158/2159-8290.CD-21-1059

11. Boccia S, Pastorino R, Ricciardi W, Ádány R, Barnhoorn F, Boffetta P, et al. How to Integrate Personalized Medicine into Prevention? Recommendations from the Personalized Prevention of Chronic Diseases (PRECeDI) Consortium. *Public Health Genomics*. 2019;22(5-6):208-214. doi:10.1159/000504652

12. Vrinten C, McGregor LM, Heinrich M, von Wagner C, Waller J, Wardle J, et al. What do people fear about cancer? A systematic review and meta-synthesis of cancer fears in the general population. *Psychooncology*. 2017;26(8):1070-1079. doi:10.1002/PON.4287

13. Vaccarella S, Georges D, Bray F, Ginsburg O, Charvat H, Martikainen P, et al. Socioeconomic inequalities in cancer mortality between and within countries in Europe: a population-based study. *Lancet Reg Heal - Eur*. 2023;25.

doi:10.1016/j.lanepe.2022.100551

14. Zhao J, Xu L, Sun J, Song M, Wang L, Yuan S, et al. Global trends in incidence, death, burden and risk factors of early-onset cancer from 1990 to 2019. *BMJ Oncol*. 2023;2(1):e000049. doi:10.1136/BMJONC-2023-000049

15. Marcus PM. *Assessment of cancer screening: a primer* [Internet]. Bethesda (MD): National Cancer Institute (US); 2019 Nov. Chapter 5, Population measures: cancer screening's impact. Published online 2019. Accessed December 23, 2023. https://www.ncbi.nlm.nih.gov/books/NBK550216/

16. Ugai T, Sasamoto N, Lee HY, Ando M, Song M, Tamimi RM, et al. Is early-onset cancer an emerging global epidemic? Current evidence and future implications. *Nat Rev Clin Oncol 2022 1910*. 2022;19(10):656-673. doi:10.1038/s41571-022-00672-8

17. Hanahan D, Weinberg RA. The hallmarks of cancer. *Cell*. 2000;100(1):57-70. doi:10.1016/S0092-8674(00)81683-9

18. Wu S, Zhu W, Thompson P, Hannun YA. Evaluating intrinsic and non-intrinsic cancer risk factors. *Nat Commun*. 2018;9(1):3490. doi:10.1038/s41467-018-05467-z

19. Imyanitov EN, Kuligina ES, Sokolenko AP, Suspitsin EN, Yanus GA, Iyevleva AG, et al. Hereditary cancer syndromes. *World J Clin Oncol*. 2023;14(2):40. doi:10.5306/WJCO.V14.I2.40

20. Garutti M, Foffano L, Mazzeo R, Michelotti A, Da Ros L, Viel A, et al. Hereditary Cancer Syndromes: A Comprehensive Review with a Visual Tool. *Genes (Basel)*. 2023;14(5):1025. doi:10.3390/GENES14051025/S1

21. Himes DO, Shuman HB. Hereditary Cancer Syndrome Recognition and Testing: Beyond BRCA. *J Nurse Pract*. 2020;16(7):517-522. doi:10.1016/J.NURPRA.2020.03.015

22. Tsaousis GN, Papadopoulou E, Apessos A, Agiannitopoulos K, Pepe G, Kampouri S, et al. Analysis of hereditary cancer syndromes by using a panel of genes: Novel and multiple pathogenic mutations. *BMC Cancer*. 2019;19(1):1-19. doi:10.1186/S12885-019-5756-4/FIGURES/6

23. Hall MJ, Obeid EI, Schwartz SC, Mantia-Smaldone G, Forman AD, Daly MB. Genetic testing for hereditary cancer predisposition: BRCA1/2, Lynch syndrome, and beyond. *Gynecol Oncol*. 2016;140(3):565. doi:10.1016/J.YGYNO.2016.01.019

24. Gomy I, Estevez Diz MDP. Hereditary cancer risk assessment: Essential tools for a better approach. *Hered Cancer Clin Pract*. 2013;11(1):1-8. doi:10.1186/1897-4287-11-16/

25. Price KS, Svenson A, King E, Ready K, Lazarin GA. Inherited Cancer in the Age of Next-Generation Sequencing. *Biol Res Nurs*. 2018;20(2):192-204. doi:10.1177/1099800417750746/ASSET/IMAGES/LARGE/10.1177_1099800417750746-FIG2.JPEG

26. Hébert J, Bergeron A-S, Veillette A-M, Bouchard K, Nabi H, Dorval M. Issues associated with a hereditary risk of cancer: Knowledge, attitudes and practices of nurses in oncology settings. *Can Oncol Nurs J*. 2022;32(2):272-285. doi:10.5737/23688076322272285

27. Buchanan AH, Lester Kirchner H, Schwartz MLB, Kelly MA, Schmidlen T, Jones LK, et al. Clinical outcomes of a genomic screening program for actionable genetic conditions. *Genet Med*. 2020;22(11):1874-1882. doi:10.1038/s41436-020-0876-4

28. Mucci LA, Hjelmborg JB, Harris JR, Czene K, Havelick DJ, Scheike T, et al. Familial Risk and Heritability of Cancer Among Twins in Nordic Countries. *JAMA*. 2016;315(1):68-76. doi:10.1001/JAMA.2015.17703

29. Stjepanovic N, Moreira L, Carneiro F, Balaguer F, Cervantes A, Balmaña J, et al. Hereditary gastrointestinal cancers: ESMO Clinical Practice Guidelines for diagnosis, treatment and follow-up. *Ann Oncol*. 2019;30(10):1558-1571. doi:10.1093/annonc/mdz233

30. Sessa C, Balmaña J, Bober SL, Cardoso MJ, Colombo N, Curigliano G, et al. Risk reduction and screening of cancer in hereditary breast-ovarian cancer syndromes: ESMO Clinical Practice Guideline. *Ann Oncol*. 2023;34(1):33-47. doi:10.1016

31. Daly MB, Pal T, Berry MP, Buys SS, Dickson P, Domchek SM, et al. Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic, Version 2.2021, NCCN Clinical Practice Guidelines in Oncology. *J Natl Compr Cancer Netw*. 2021;19(1):77-102. doi:10.6004/jnccn.2021.0001

32. Clinical Guideline Familial breast cancer: Classification and care of people at risk of familial breast cancer and management of breast cancer and related risks in people with a family history of breast cancer. Published online 2013. Accessed January 4, 2024. www.nice.org.uk/guidance/cg164

33. Umar A, Boland CR, Terdiman JP, Syngal S, de la Chapelle A, Rüschoff J, et al. Revised Bethesda Guidelines for Hereditary Nonpolyposis Colorectal Cancer (Lynch Syndrome) and Microsatellite Instability. *J Natl Cancer Inst*. 2004;96(4):261. doi:10.1093/JNCI/DJH034

34. Ajani JA, D'Amico TA, Bentrem DJ, Chao J, Cooke D, Corvera C, et al. Gastric Cancer, Version 2.2022, NCCN Clinical Practice Guidelines in Oncology. *J Natl Compr Cancer Netw*. 2022;20(2):167-192. doi:10.6004/JNCCN.2022.0008

35. Lee A, Mavaddat N, Wilcox AN, Cunningham AP, Carver T, Hartley S, et al. BOADICEA: a comprehensive breast cancer risk prediction model incorporating genetic and nongenetic risk factors. *Genet Med*. 2019;21(8):1708-1718. doi:10.1038/s41436-018-0406-9

36. Hart SN, Polley EC, Yussuf A, Yadav S, Goldgar DE, Hu C, et al. Mutation prevalence tables for hereditary cancer derived from multigene panel testing. *Hum Mutat*. 2020;41(8):e1. doi:10.1002/HUMU.24053

37. Heald B, Marquard J, Funchain P. Strategies for clinical implementation of screening for hereditary cancer syndromes. *Semin Oncol*. 2016;43(5):609-614. doi:10.1053/J.SEMINONCOL.2016.08.008

38. Bhattacharya P, McHugh TW. Lynch Syndrome. *Encycl Gastroenterol Second Ed*. Published online February 4, 2023:490-494. doi:10.1016/B978-0-12-801238-3.65755-0

39. B Beitsch PD, Whitworth PW, Hughes K, Patel R, Rosen B, Compagnoni G, et al. Underdiagnosis of Hereditary Breast Cancer: Are Genetic Testing Guidelines a Tool or an Obstacle? *J Clin Oncol*. 2019;37(6):453-460. doi:10.1200/JCO.18.01631

40. Gallon R, Gawthorpe P, Phelps RL, Hayes C, Borthwick GM, Santibanez-Koref M, et al. How Should We Test for Lynch Syndrome? A Review of Current Guidelines and Future Strategies. *Cancers (Basel)*. 2021;13(3):406. Accessed January 4, 2024. https://www.mdpi.com/2072-6694/13/3/406/htm

41. Laforest F, Mann B, Edwards A, Kirkegaard P. Genetic cancer risk assessment in general practice: Systematic review of tools available, clinician attitudes, and patient outcomes. *Br J Gen Pract*. 2019;69(679):E97-E105. doi:10.3399/bjgp18X700265

42. Grzymski JJ, Elhanan G, Morales Rosado JA, Smith E, Schlauch KA, Read R, et al. Population genetic screening efficiently identifies carriers of autosomal dominant diseases. *Nat Med 2020 268*. 2020;26(8):1235-1239. doi:10.1038/s41591-020-0982-5

43. Riley BD, Culver JO, Skrzynia C, Senter LA, Peters JA, Costalas JW, et al. Essential elements of genetic cancer risk assessment, counseling, and testing: Updated recommendations of the National Society of genetic Counselors. *J Genet Couns*. 2012;21(2):151-161. doi:10.1007/s10897-011-9462-x

44. Martínez MT, Tapia M, Candia L, Bermejo B, Garrido-Cano I, Adam-Artigues A, et al. Hereditary Risk Assessment for BRCA Breast and/or Ovarian Cancer. *J Cancer Sci Clin Ther*. 2022;06(03). doi:10.26502/jcsct.5079162

45. Colibaba A, Azoicai D, Gheorghiu I, Matei M. The HOPE project: New initiatives in the detection and prevention of hereditary cancers. In: *2019 7th E-Health and Bioengineering Conference, EHB 2019*. Institute of Electrical and Electronics Engineers Inc.; 2019. doi:10.1109/EHB47216.2019.8970037

46. Wilson JMG, Jungner G. *Principes and Practice of Screening for Disease*. World Health Organisation; 1968.

47. Hampel H. Genetic counseling and cascade genetic testing in Lynch syndrome. *Fam Cancer*. 2016;15(3):423-427. doi:10.1007/s10689-016-9893-5

48. Desmond A, Kurian AW, Gabree M, Mills MA, Anderson MJ, Kobayashi Y, et al. Clinical actionability of multigene panel testing for hereditary breast and ovarian cancer risk assessment. *JAMA Oncol*. 2015;1(7):943-951. doi:10.1001/jamaoncol.2015.2690

49. Cragun D, Beckstead J, Farmer M, Hooker G, Dean M, Matloff E, et al. IMProving care After inherited Cancer Testing (IMPACT) study: protocol of a randomized trial evaluating the efficacy of two interventions designed to improve cancer risk management and family communication of genetic test results. *BMC Cancer*. 2021;21(1):1099. doi:10.1186/s12885-021-08822-4

50. Evans DGR, Ingham SL. Reduced life expectancy seen in hereditary diseases which predispose to early-onset tumors. *Appl Clin Genet*. 2013;6:53. doi:10.2147/TACG.S35605

51. Mittendorf KF, Hunter JE, Schneider JL, Shuster E, Rope AF, Zepp J, et al. Recommended care and care adherence following a diagnosis of Lynch syndrome: A mixed-methods study. *Hered Cancer Clin Pract*. 2019;17(1). doi:10.1186/s13053-019-0130-8

52. K Kratz CP, Achatz MI, Brugieres L, Frebourg T, Garber JE, Greer MLC, et al. Cancer screening recommendations for individuals with Li-Fraumeni syndrome. *Clin Cancer Res*. 2017;23(11):e38-e45. doi:10.1158/1078-0432.CCR-17-0408

53. Paul JL, Leslie H, Trainer AH, Gaff C. A theory-informed systematic review of clinicians' genetic testing practices. *Eur J Hum Genet*. 2018;26(10):1401. doi:10.1038/S41431-018-0190-7

54. Abacan MA, Alsubaie L, Barlow-Stewart K, Caanen B, Cordier C, Courtney E, et al. The Global State of the Genetic Counseling Profession. *Eur J Hum Genet*. 2019;27(2):183-197. doi:10.1038/s41431-018-0252-x

55. Resta RG. What have we been trying to do and have we been any good at it? A history of measuring the success of genetic counseling. *Eur J Med Genet*. 2019;62(5):300-307. doi:10.1016/j.ejmg.2018.11.003

56. Madlensky L, Trepanier AM, Cragun D, Lerner B, Shannon KM, Zierhut H. A Rapid Systematic Review of Outcomes Studies in Genetic Counseling. *J Genet Couns*. 2017;26(3):361-378. doi:10.1007/S10897-017-0067-X

57. Athens BA, Caldwell SL, Umstead KL, Connors PD, Brenna E, Biesecker BB. A Systematic Review of Randomized Controlled Trials to Assess Outcomes of Genetic Counseling. *J Genet Couns*. 2017;26(5):902-933. doi:10.1007/S10897-017-0082-Y

58. McCuaig JM, Armel SR, Care M, Volenik A, Kim RH, Metcalfe KA. Next-Generation Service Delivery: A Scoping Review of Patient Outcomes Associated with Alternative Models of Genetic Counseling and Genetic Testing for Hereditary Cancer. *Cancers (Basel)*. 2018;10(11):435. doi:10.3390/cancers10110435

59. Eggert J. Genetics and Genomics in Oncology Nursing. *Nurs Clin North Am*. 2017;52(1):1-25. doi:10.1016/j.cnur.2016.11.001

60. Schneider JL, Goddard KAB, Muessig KR, Davis J V., Rope AF, Hunter JE, et al. Patient and provider perspectives on adherence to and care coordination of lynch syndrome surveillance recommendations: Findings from qualitative interviews. *Hered Cancer Clin Pract*. 2018;16(1). doi:10.1186/s13053-018-0090-4

61. Pollard S, Kalloger S, Weymann D, Sun S, Nuk J, Schrader KAKA, et al. Genetic testing for hereditary cancer syndromes: patient recommendations for improved risk communication. *Heal Expect*. 2020;23(4):884-892. doi:10.1111/hex.13062

62. Singer CF, Balmaña J, Bürki N, Delaloge S, Filieri ME, Gerdes AM, et al.Genetic counselling and testing of susceptibility genes for therapeutic decision-making in breast cancer—an European consensus statement and expert recommendations. *Eur J Cancer*. 2019;106:54-60. doi:10.1016/j.ejca.2018.10.007

63. Dean M. "It's not if I get cancer, it's when I get cancer": BRCA-positive patients' (un)certain health experiences regarding hereditary breast and ovarian cancer risk. *Soc Sci Med*. 2016;163:21-27. doi:10.1016/J.SOCSCIMED.2016.06.039

64. Campbell-Salome G, Buchanan AH, Hallquist MLG, Rahm AK, Rocha H, Sturm AC. Uncertainty management for individuals with Lynch Syndrome: Identifying and responding to healthcare barriers. *Patient Educ Couns*. 2021;104(2):403-412. doi:10.1016/J.PEC.2020.07.017

65. Warner NZ, Groarke AM. A qualitative reflexive thematic analysis into the experiences of being identified with a BRCA1/2 gene alteration: "So many little, little traumas could have been avoided." *BMC Health Serv Res*. 2022;22(1):1-12. doi:10.1186/S12913-022-08372-W/TABLES/2

66. Mooney R, Wu YP, Kehoe K, Volkmar M, Kohlmann W, Koptiuch C, et al. Experiences of patients and family members with follow-up care, information needs and provider support after identification of Lynch Syndrome. *Hered Cancer Clin Pract*. 2023;21(1):1-9. doi:10.1186/S13053-023-00273-1

67. Stan DL, Shuster LT, Wick MJ, Swanson CL, Pruthi S, Bakkum-Gamez JN. Challenging and complex decisions in the management of the BRCA mutation carrier. *J Women's Heal*. 2013;22(10):825-834. doi:10.1089/jwh.2013.4407

68. Torrisi C. Body Image in BRCA-Positive Young Women Following Bilateral Risk-Reducing Mastectomy: A Review of the Literature. *Front Psychol*. 2021;12:778484. doi:10.3389/FPSYG.2021.778484/BIBTEX

69. Gomes P, Pietrabissa G, Silva ERE, Silva J, Matos PM, Costa ME, et al. Family Adjustment to Hereditary Cancer Syndromes: A Systematic Review. *Int J Environ Res Public Health*. 2022;19(3):1603. Accessed June 28, 2022. https://pubmed.ncbi.nlm.nih.gov/35162625/

70. Metcalfe A, Plumridge G, Coad J, Shanks A, Gill P. Parents' and children's communication about genetic risk: a qualitative study, learning from families' experiences. *Eur J Hum Genet*. 2011;19(6):640-646. doi:10.1038/ejhg.2010.258

71. Santos EMM, Edwards QT, Floria-Santos M, Rogatto SR, Achatz MIW, MacDonald DJ. Integration of Genomics in Cancer Care. *J Nurs Scholarsh*. 2013;45(1):43-51. doi:10.1111/j.1547-5069.2012.01465.x

72. Hynes J, Dawson L, Seal M, Green J, Woods M, Etchegary H. "There should be one spot that you can go:" BRCA mutation carriers' perspectives on cancer risk management and a hereditary cancer registry. *J Community Genet*. 2023;1:1-10. doi:10.1007/S12687-023-00685-5/TABLES/3

73. Dibble KE, Donorfio LKM, Britner PA, Bellizzi KM. Perceptions and care Recommendations from Previvors: Qualitative analysis of female BRCA1/2 mutation Carriers' experience with genetic testing and counseling. *Gynecol Oncol Reports*. 2022;41:100989. doi:10.1016/J.GORE.2022.100989

74. Carvalho F, Koziel A, Monje-Garcia L. How to set up an in-house nurse-led colorectal Lynch syndrome clinic. *Gastrointest Nurs*. 2023;21(6):22-25. doi:10.12968/gasn.2023.21.6.22

75. Dean M, Scherr CL, Clements M, Koruo R, Martinez J, Ross A. "When information is not enough": A model for understanding BRCA-positive previvors' information needs regarding hereditary breast and ovarian cancer risk. *Patient Educ Couns*. 2017;100(9):1738-1743. doi:10.1016/J.PEC.2017.03.013

76. Williams JK, Katapodi MC, Starkweather A, Badzek L, Cashion AK, Coleman B, et al. Advanced nursing practice and research contributions to precision medicine. *Nurs Outlook*. 2016;64(2):117-123. doi:10.1016/j.outlook.2015.11.009

77. Young AM, Charalambous A, Owen RI, Njodzeka B, Oldenmenger WH, Alqudimat MR, et al. Essential oncology nursing care along the cancer continuum. *Lancet Oncol*. 2020;21(12):e555-e563. doi:10.1016/S1470-2045(20)30612-4

78. Charalambous A, Wells M, Campbell P, Torrens C, Östlund U, Oldenmenger W, et al. A scoping review of trials of interventions led or delivered by cancer nurses. *Int J Nurs Stud*. 2018;86:36-43. doi:10.1016/J.IJNURSTU.2018.05.014

79. Calzone KA, Cashion A, Feetham S, Jenkins J, Prows CA, Williams JK, et al. Nurses transforming health care using genetics and genomics. *Nurs Outlook*. 2010;58(1):26-35. doi:10.1016/J.OUTLOOK.2009.05.001

80. Hyatt A, Shelly A, Cox R, Humphries E, Lock G, Varlow M. How can we improve information for people affected by cancer? A national survey exploring gaps in current information provision, and challenges with accessing cancer information online. *Patient Educ Couns*. 2022;105(8):2763-2770. doi:10.1016/J.PEC.2022.04.009

81. Hann KEJ, Freeman M, Fraser L, Waller J, Sanderson SC, Rahman B, et al. Awareness,

knowledge, perceptions, and attitudes towards genetic testing for cancer risk among ethnic minority groups: A systematic review. *BMC Public Health*. 2017;17(1). doi:10.1186/s12889-017-4375-8

82. Starkings R, Shilling V, Jenkins V, Fallowfield L. A systematic review of communication interventions to help healthcare professionals discuss genetic testing for breast cancer. *Breast Cancer Res Treat*. 2020;183(1):9. doi:10.1007/S10549-020-05741-Z

83. Peterson EB, Chou WYS, Gaysynsky A, Krakow M, Elrick A, Khoury MJ, et al. Communication of Cancer-related genetic and genomic information: A landscape analysis of reviews. *Transl Behav Med*. 2018;8(1):59-70. doi:10.1093/tbm/ibx063

84. Papaioannou K, Kampourakis K. Health-Care Professionals' Awareness and Understanding of Genomics. *Appl Genomics Public Heal*. Published online January 1, 2020:225-242. doi:10.1016/B978-0-12-813695-9.00012-1

85. Calzone KA, Kirk M, Tonkin E, Badzek L, Benjamin C, Middleton A. Increasing nursing capacity in genomics: Overview of existing globalgenomics resources. *Nurse Educ Today*. 2018;69:53. doi:10.1016/J.NEDT.2018.06.032

86. Matsumoto M, Sasaki N, Tsukigawa Y, Otsubo R, Yano H, Nagayasu T. A Survey of the Awareness and Educational Needs of Nurses in Nagasaki Prefecture Regarding Hereditary Breast and Ovarian Cancer. *J Cancer Educ*. Published online January 6, 2022:1-6. doi:10.1007/S13187-022-02132-4/TABLES/2

87. Jacobs C, Rahman B. One size does not fit all: The case for targeted education in genetics and genomics for cancer nurses. *Eur J Cancer Care (Engl)*. 2021;30(4):e13480. doi:10.1111/ECC.13480

88. Wild CP, Weiderpass E, Stewart BW. World Cancer Report: Cancer Research for Cancer Prevention. *International Agency for Research on Cancer*, 2020. Accessed October 2, 2021. http://gco.iarc.fr/today

89. Clinton SK, Giovannucci EL, Hursting SD. The World Cancer Research Fund/American Institute for Cancer Research Third Expert Report on Diet, Nutrition, Physical Activity, and Cancer: Impact and Future Directions. *J Nutr*. 2020;150(4):663-671. doi:10.1093/JN/NXZ268

90. International Agency for Research on Cancer. *IARC Handbooks of Cancer Prevention: Preamble – Primary Prevention*. IARC; 2019.

91. Safiri S, Nejadghaderi SA, Abdollahi M, Carson-Chahhoud K, Kaufman JS, Bragazzi NL, et al. Global, regional, and national burden of cancers attributable to tobacco smoking in 204 countries and territories, 1990–2019. *Cancer Med*. 2022;11(13):2662. doi:10.1002/CAM4.4647

92. Pati S, Irfan W, Jameel A, Ahmed S, Shahid RK. Obesity and Cancer: A Current Overview of Epidemiology, Pathogenesis, Outcomes, and Management. *Cancers (Basel)*. 2023;15(2). doi:10.3390/CANCERS15020485

93. Rumgay H, Lam F, Ervik M, Soerjomataram I. Cancers attributable to alcohol. Lyon, France: International Agency for Research on Cancer. Published 2021. Accessed July 20, 2021. https://gco.iarc.fr/causes/alcohol, accessed [20 July 2021

94. Lammert J, Grill S, Kiechle M. Modifiable Lifestyle Factors: Opportunities for (Hereditary) Breast Cancer Prevention - A Narrative Review. *Breast Care*. 2018;13(2):109-114. doi:10.1159/000488995

95. Schüz J, Espina C, Villain P, Herrero R, Leon ME, Minozzi S, et al. European Code against Cancer 4th Edition: 12 ways to reduce your cancer risk. *Cancer Epidemiol*. 2015;39:S1-S10. doi:10.1016/J.CANEP.2015.05.009

96. World Cancer Research Fund International. Our Cancer Prevention Recommendations. WCRF. Published 2016. Accessed July 20, 2021. https://www.wcrf.org/diet-and-cancer/cancer-prevention-recommendations/

97. Quillin JM. Lifestyle Risk Factors Among People Who Have Had Cancer Genetic Testing. *J Genet Couns*. 2016;25(5):957-964. doi:10.1007/s10897-015-9925-6

98. Cho YA, Lee J, Oh JH, Chang HJ, Sohn DK, Shin A, et al. Genetic risk score, combined lifestyle factors and risk of colorectal cancer. *Cancer Res Treat*. 2019;51(3):1033-1040. doi:10.4143/crt.2018.447

99. Lazzeroni M, Bellerba F, Calvello M, Macrae F, Win A, Jenkins M, et al. A Meta-Analysis of Obesity and Risk of Colorectal Cancer in Patients with Lynch Syndrome: The Impact of Sex and Genetics. *Nutrients*. 2021;13(5). doi:10.3390/NU13051736

100. Deng N, Reyes-Uribe L, Fahrmann JF, Thoman WS, Munsell MF, Dennison JB, et al. Exercise Training Reduces the Inflammatory Response and Promotes Intestinal Mucosa-Associated Immunity in Lynch Syndrome. *Clin Cancer Res*. 2023;29(21):4361-4372. doi:10.1158/1078-0432.CCR-23-0088/729051/AM/EXERCISE-TRAINING-REDUCES-THE-INFLAMMATORY

101. Coletta AM, Peterson SK, Gatus LA, Krause KJ, Schembre SM, Gilchrist SC, et al. Diet, weight management, physical activity and Ovarian & Breast Cancer Risk in women with BRCA1/2 pathogenic Germline gene variants: Systematic review. *Hered Cancer Clin Pract*. 2020;18(1):1-24. doi:10.1186/s13053-020-0137-1

102. Daniele A, Divella R, Pilato B, Tommasi S, Pasanisi P, Patruno M, et al. Can harmful lifestyle, obesity and weight changes increase the risk of breast cancer in BRCA 1 and BRCA 2 mutation carriers? A Mini review. *Hered Cancer Clin Pract*. 2021;19(1):1-8. doi:10.1186/S13053-021-00199-6

103. Al Ajmi K, Lophatananon A, Mekli K, Ollier W, Muir KR. Association of Nongenetic Factors With Breast Cancer Risk in Genetically Predisposed Groups of Women in the UK Biobank Cohort. *JAMA Netw open*. 2020;3(4):e203760. doi:10.1001/jamanetworkopen.2020.3760

104. Visser A, Vrieling A, Murugesu L, Hoogerbrugge N, Kampman E, Hoedjes M. Determinants of adherence to recommendations for cancer prevention among Lynch Syndrome mutation carriers: A qualitative exploration. *PLoS One*. 2017;12(6):1-12. doi:10.1371/journal.pone.0178205

105. Mazor KM, Roblin DW, Williams AE, Greene SM, Gaglio B, Field TS, et al. Health Literacy and Cancer Prevention: Two New Instruments to Assess Comprehension. *Patient Educ Couns*. 2012;88(1):54. doi:10.1016/J.PEC.2011.12.009

106. Hollands GJ, French DP, Griffin SJ, Prevost AT, Sutton S, King S, et al. The impact of communicating genetic risks of disease on riskreducing health behaviour: Systematic review with meta-analysis. *BMJ*. 2016;352. doi:10.1136/bmj.i1102

107. Wang W, Fang S, Zhang S, He M, Zhu X, Dong Y, et al. Gaps in awareness and practice of healthy lifestyles among individuals at high risk of colorectal cancer: A qualitative evidence synthesis. *J Clin Nurs*. 2023;32(17-18):5737-5751. doi:10.1111/JOCN.16696

108. Anderson AS, Caswell S, Macleod M, Steele RJ, Berg J, Dunlop J, et al. Health Behaviors and their Relationship with Disease Control in People Attending Genetic Clinics with a Family History of Breast or Colorectal Cancer. *J Genet Couns*. 2017;26(1):40-51. doi:10.1007/s10897-016-9977-2

109. Stevens C, Vrinten C, Smith SG, Waller J, Beeken RJ. Acceptability of receiving lifestyle advice at cervical, breast and bowel cancer screening. *Prev Med (Baltim)*. 2019;120(November 2018):19-25. doi:10.1016/j.ypmed.2018.12.005

110. Hibbard J, Gilburt H. Supporting people to manage their health: an introduction to patient activation. King's Fund. 2014. United Kingdom. Published online 2014. Accessed January 15, 2024. https://policycommons.net/artifacts/1716007/supporting-people-to-manage-their-health/2447678/

111. Orange ST, Hicks KM, Saxton JM. Effectiveness of diet and physical activity interventions amongst adults attending colorectal and breast cancer screening: a systematic review and meta-analysis. *Cancer Causes Control*. Published online November 8, 2020:1-14. doi:10.1007/s10552-020-01362-5

112. Spees CK, Braun AC, Hill EB, Grainger EM, Portner J, Young GS, et al. Impact of a Tailored Nutrition and Lifestyle Intervention for Overweight Cancer Survivors on Dietary Patterns, Physical Activity, Quality of Life, and Cardiometabolic Profiles. *J Oncol*. 2019;2019:1-13. doi:10.1155/2019/1503195

113. Vrieling A, Visser A, Hoedjes M, Hurks M, Gómez García E, Hoogerbrugge N, et al. Increasing awareness and knowledge of lifestyle recommendations for cancer prevention in Lynch syndrome carriers: Randomized controlled trial. *Clin Genet*. 2018;93(1):67-77. doi:10.1111/cge.13076

114. Sinclair J, McCann M, Sheldon E, Gordon I, Brierley-Jones L, Copson E. The acceptability of addressing alcohol consumption as a modifiable risk factor for breast cancer: A mixed method study within breast screening services and symptomatic breast clinics. *BMJ Open*. 2019;9(6):1-12. doi:10.1136/bmjopen-2018-027371

115. Underhill-Blazey M, Blonquist T, Lawrence J, Hong F, Yurgelun MB, Syngal S. Health behaviours and beliefs in individuals with familial pancreatic cancer. *Fam Cancer*. 2019;18(4):457-464. doi:10.1007/s10689-019-00143-7

116. Michie S, Atkins L, West R. *The Behaviour Change Wheel. A Guide to Designing Interventions.* 1st Ed. Silverback Publishing; 2016.

117. Tlusty K, Jackson M, Riley B, Blase T. Effects of diet education on empowerment for individuals who have an increased risk of developing breast or colon cancer: A pilot study. *J Genet Couns*. 2022;31(5):1138-1147. doi:10.1002/JGC4.1584

118. Anderson AS, Dunlop J, Gallant S, Macleod M, Miedzybrodzka Z, Mutrie N, et al. Feasibility study to assess the impact of a lifestyle intervention (a € LivingWELL') in people having an assessment of their family history of colorectal or breast cancer. *BMJ Open*. 2018;8(2):1-11. doi:10.1136/bmjopen-2017-019410

119. Edwards AGK, Naik G, Ahmed H, Elwyn GJ, Pickles T, Hood K, et al. Personalised risk communication for informed decision making about taking screening tests. *Cochrane Database Syst Rev*. 2013;2013(2):1-96. doi:10.1002/14651858.CD001865.pub3

120. Mittendorf KF, Knerr S, Kauffman TL, Lindberg NM, Anderson KP, Feigelson HS, et al. Systemic Barriers to Risk-Reducing Interventions for Hereditary Cancer Syndromes:

Implications for Health Care Inequities. *JCO Precis Oncol*. 2021;(5):1709-1718. doi:10.1200/PO.21.00233

121. Hickey KT, Taylor JY, Barr TL, Hauser NR, Jia H, Riga TC, et al. Nursing genetics and genomics: The International Society of Nurses in Genetics (ISONG) survey. *Nurse Educ Today*. 2018;63(January):12-17. doi:10.1016/j.nedt.2018.01.002

122. Mahon SM, Yackzan S. Oncology Nurse Practitioners in Genetics: Examining Scope of Practice and Competence. *Clin J Oncol Nurs*. 2022;26(2):141-145. doi:10.1188/22.CJON.141-145

123. Glanz K, Rimer B k., Viswanath K. *Health Behavior and Health Education: Theory, Research, and Practice*. 4th ed. (Glanz K, Rimer B k., Viswanath K, eds.). Jossey-Bass; 2008. Accessed May 14, 2022. https://www.academia.edu/download/49289960/Health_Behavior___Health_Education_book_4th_Ed.pdf#page=83

124. Patterson BJ. A Dynamic Relationship Between Theory and Research in Nursing Education. *Nurs Educ Perspect*. 2021;42(6):337-338. doi:10.1097/01.NEP.0000000000000903

125. Anwar Tayel A, Hosny H, Sherbini E, Abd A, Fattah E, Shosha A, et al. The Effect of Applying Preventive Intervention Based on Champion Health Believe Model on Breast Cancer Fatalism, Knowledge and Screening Behaviors among Female Employees. *Am J Nurs Res*. 2019;7(5):759-770. doi:10.12691/ajnr-7-5-9

126. Orji R, Vassileva J, Mandryk R. Towards an Effective Health Interventions Design: An Extension of the Health Belief Model. *Online J Public Health Inform*. 2012;4(3). doi:10.5210/OJPHI.V4I3.4321

127. Rakhshanderou S, Maghsoudloo M, Safari-Moradabadi A, Ghaffari M. Theoretically designed interventions for colorectal cancer prevention: A case of the health belief model. *BMC Med Educ*. 2020;20(1):1-8. doi:10.1186/S12909-020-02192-4/FIGURES/3

128. Lau J, Lim TZ, Jianlin Wong G, Tan KK. The health belief model and colorectal cancer screening in the general population: A systematic review. *Prev Med Reports*. 2020;20:101223. doi:10.1016/J.PMEDR.2020.101223

129. Fleary SA, Paasche-Orlow MK, Joseph P, Freund KM. The Relationship Between Health Literacy, Cancer Prevention Beliefs, and Cancer Prevention Behaviors. *J Cancer Educ*. 2019;34(5):958-965. doi:10.1007/S13187-018-1400-2

130. Buchanan AH, Voils CI, Schildkraut JM, Fine C, Horick NK, Marcom PK, et al. Adherence to Recommended Risk Management among Unaffected Women with a BRCA Mutation. *J Genet Couns*. 2017;26(1):79-92. doi:10.1007/s10897-016-9981-6

131. Ydalsys Naranjo Hernández C, José C, Pacheco AC, Miriam L, Larreynaga R. The self-care deficit nursing theory: Dorothea Elizabeth Orem. 2017;19(3).

132. Alligood MR. Modelos y teorías en enfermería. Published online 2022. Accessed January 20, 2024. https://www.casadellibro.com/libro-modelos-y-teorias-en-enfermeria-10-ed/9788413822990/13224368

133. Rakhshani T, Najafi S, Javady F, Taghian dasht bozorg A, Mohammadkhah F, Khani Jeihooni A. The effect of Orem-based self-care education on improving self-care ability of patients undergoing chemotherapy: a randomized clinical trial. *BMC Cancer*. 2022;22(1):1-9. doi:10.1186/S12885-022-09881-X/TABLES/5

134. RobatSarpooshi D, Mahdizadeh M, Alizadeh Siuki H, Haddadi M, Robatsarpooshi H, Peyman N. The Relationship Between Health Literacy Level and Self-Care Behaviors in Patients with Diabetes. *Patient Relat Outcome Meas*. 2020;Volume 11:129-135. doi:10.2147/PROM.S243678

135. Setiawan H, Suhanda S, Roslianti E, Firmansyah A. Theory Development of Genetic Counseling among Patient with Genetic Diseases. *Int J Nurs Heal Serv*. 2020;3(6):709-715. doi:10.35654/IJNHS.V3I6.350

136. Jahoda M. Current concepts of positive mental health. *Curr concepts Posit Ment Heal*. Published online December 13, 2006. doi:10.1037/11258-000

137. Lluch Canut MT. Construcción de una escala para evaluar la salud mental positiva. Doctoral thesis. Faculty of Psychology, University of Barcelona, Barcelona, Spain.1999. Published online 2004. Accessed October 22, 2021. http://www.tdx.cat/TDX-03311 04-104403

138. Lluch-Canut T, Puig-Llobet M, Sánchez-Ortega A, Roldán-Merino J, Ferré-Grau C, Albacar N, et al. Assessing positive mental health in people with chronic physical health problems: Correlations with socio-demographic variables and physical health status. *BMC Public Health*. 2013;13(1):928. doi:10.1186/1471-2458-13-928

139. Sánchez-Ortega MA, Lluch-Canut MT, Roldán-Merino J, Agüera Z, Hidalgo-Blanco MA, Moreno-Poyato AR, et al. Nursing Intervention to Improve Positive Mental Health and Self-Care Skills in People with Chronic Physical Health Conditions. *Int J Environ Res Public Health*. 2022;20(1):528. doi:10.3390/ijerph20010528

140. Puig Llobet M, Sánchez Ortega M, Lluch-Canut M, Moreno-Arroyo M, Hidalgo Blanco MÀ, Roldán-Merino J. Positive Mental Health and Self-Care in Patients with Chronic Physical Health Problems: Implications for Evidence-based Practice. *Worldviews Evidence-Based Nurs*. 2020;17(4):293-300. doi:10.1111/WVN.12453

141. Holtmaat K, van der Spek N, Lissenberg-Witte BI, Cuijpers P, Verdonck-de Leeuw IM. Positive mental health among cancer survivors: overlap in psychological well-being, personal meaning, and posttraumatic growth. *Support Care Cancer*. 2019;27(2):443. doi:10.1007/S00520-018-4325-8

142. Ma Y, Xie T, Zhang J, Yang H. The prevalence, related factors and interventions of oncology nurses' burnout in different continents: A systematic review and meta-analysis. *J Clin Nurs*. 2023;32(19-20):7050-7061. doi:10.1111/JOCN.16838

143. Benito L, Binefa G, Vidal C, Lluch MT, Puig M, Padrol I, et al. Identifying nursing activities in population-based colorectal and breast cancer screening programs in Spain: A Delphi study. *Collegian*. 2017;24(4):351-359. doi:10.1016/J.COLEGN.2016.07.005

144. Fernández Guijarro S, Pomarol-Clotet E, Rubio Muñoz MC, Miguel García C, Egea López E, Fernández Guijarro R, et al. Effectiveness of a community-based nurse-led lifestyle-modification intervention for people with serious mental illness and metabolic syndrome. *Int J Ment Health Nurs*. 2019;28(6):1328-1337. doi:10.1111/inm.12644

145. French DP, Cameron E, Benton JS, Deaton C, Harvie M. Can Communicating Personalised Disease Risk Promote Healthy Behaviour Change? A Systematic Review of Systematic Reviews. *Ann Behav Med*. 2017;51(5):718-729. doi:10.1007/s12160-017-9895-z

146. Jenkins CD. *Building a Better Health. A Handbook of Behavioural Change*. World Health

Organization; 2003.

147. Roberts L, Turner K, George' S, Ward D. Briefing notes for researchers - public involvement in NHS, health and social care research. NIHR. Published online 2012. Accessed July 27, 2023. https://www.nihr.ac.uk/documents/briefing-notes-for-researchers-public-involvement-in-nhs-health-and-social-care-research/273711.

148. National Institute for Health Research (NIHR)-wide learning and development for public involvement: working group report and recommendations. Published online 2015.

149. Hoffmann TC, Glasziou PP, Boutron I, Milne R, Perera R, Moher D, et al. Better reporting of interventions: template for intervention description and replication (TIDieR) checklist and guide. *BMJ*. 2014;348. doi:10.1136/BMJ.G1687

150. Tufanaru C, Munn Z, Aromataris E, Campbell J, Hopp L. Chapter 3: Systematic Reviews of Effectiveness. *JBI Man Evid Synth*. Published online 2020. doi:10.46658/JBIMES-20-04

151. Moher D, Liberati A, Tetzlaff J, Altman DG. *Preferred Reporting Items for Systematic Reviews and Meta-Analyses: The PRISMA Statement*. Vol 339. British Medical Journal Publishing Group; 2009:332-336. doi:10.1136/bmj.b2535

152. Okoli C, Pawlowski SD. The Delphi method as a research tool: an example, design considerations and applications. *Inf Manag*. 2004;42(1):15-29. doi:10.1016/j.im.2003.11.002

153. Shariff NJ. Utilizing the Delphi Survey Approach: A Review. *J Nurs Care*. 2015;4(3):246-251. doi:10.4172/2167-1168.1000246

154. European Oncology Nursing Society (EONS). *The EONS Cancer Nursing Education Framework*. Accessed October 26, 2021. https://cancernurse.eu/education/cancer-nursing-education-framework/

155. Niederberger M, Spranger J. Delphi Technique in Health Sciences: A Map. *Front Public Heal*. 2020;8:457. doi:10.3389/FPUBH.2020.00457/BIBTEX

156. Willis DG, Leone-Sheehan DM. *Dorothea E. Orem: Teoría Del Défi Cit de Autocuidado*. Vol 6. 9th ed. Elsevier; 2018. Accessed January 20, 2024. https://dialnet.unirioja.es/servlet/articulo?codigo=6686834

157. Braun V, Clarke V, Hayfield N, Terry G. Thematic analysis. *Handb Res Methods Heal Soc Sci*. Published online January 12, 2019:843-860. doi:10.1007/978-981-10-5251-4_103/COVER

158. Braun V, Clarke V. One size fits all? What counts as quality practice in (reflexive) thematic analysis? *Qual Res Psychol*. 2020;18(3):328-352. doi:10.1080/14780887.2020.1769238

159. Braun V, Clarke V. To saturate or not to saturate? Questioning data saturation as a useful concept for thematic analysis and sample-size rationales. *Qual Res Sport Exerc Heal*. 2021;13(2):201-216. doi:10.1080/2159676X.2019.1704846

160. Dineen-Griffin S, Garcia-Cardenas V, Williams K, Benrimoj SI. Helping patients help themselves: A systematic review of self-management support strategies in primary health care practice. *PLoS One*. 2019;14(8). doi:10.1371/JOURNAL.PONE.0220116

161. Koutsopoulou S, Papathanassoglou ED, Katapodi MC, Patiraki EI. A critical review of the evidence for nurses as information providers to cancer patients. *J Clin Nurs*. 2010;19(5-6):749-765. doi:10.1111/j.1365-2702.2009.02954.x

162. Dinç L, Gastmans C. Trust in nurse–patient relationships. *Nurs Ethics*. 2013;20(5):501-516. doi:10.1177/0969733012468463

163. Kelly D, Lankshear A, Wiseman T, Jahn P, Mall-Roosmäe H, Rannus K, et al. The experiences of cancer nurses working in four European countries: A qualitative study. *Eur J Oncol Nurs*. 2020;49. doi:10.1016/J.EJON.2020.101844

164. Challinor JM, Alqudimat MR, Teixeira TOA, Oldenmenger WH. Oncology nursing workforce: challenges, solutions, and future strategies. *Lancet Oncol*. 2020;21(12):e564-e574. doi:10.1016/S1470-2045(20)30605-7

165. So WK, Chan RJ, Truant T, Trevatt P, Bialous SA, Barton-Burke M. Global perspectives on cancer health disparities: Impact, utility, and implications for cancer nursing. *Asia-Pacific J Oncol Nurs*. 2016;3(4):316-323. doi:10.4103/2347-5625.195885

166.. Warner NZ, Gleeson C, Fahey P, Horgan R, Groarke AM. Experiences of living with Lynch Syndrome: A reflexive thematic analysis. *Eur J Oncol Nurs*. 2022;58:102117. doi:10.1016/J.EJON.2022.102117

167. Diez de los Rios de la Serna C, Fernández-Ortega P, Lluch-Canut T. Lifestyle Behavior Interventions for Preventing Cancer in Adults with Inherited Cancer Syndromes: Systematic Review. *Int J Environ Res Public Health*. 2022;19(21). doi:10.3390/ijerph192114098

168. Anderson AS, Craigie AM, Gallant S, McAdam C, Macaskill EJ, Mutrie N, et al. Randomised controlled trial to assess the impact of a lifestyle intervention (ActWELL) in women invited to NHS breast screening. *BMJ Open*. 2018;8(11):1-11. doi:10.1136/bmjopen-2018-024136

169. Horne J, Madill J, O'Connor C, Shelley J, Gilliland J. A Systematic Review of Genetic Testing and Lifestyle Behaviour Change: Are We Using High-Quality Genetic Interventions and Considering Behaviour Change Theory? *Lifestyle Genomics*. 018;11(1):49-63. doi:10.1159/000488086

170. Cummins KM. Explanations for the Cloudy Evidence That Theory Benefits Health Promotion. *Front Psychol*. 2022;13:910041. doi:10.3389/FPSYG.2022.910041

171. Bartholomew LK, Mullen PD. Five roles for using theory and evidence in the design and testing of behavior change interventions. *J Public Health Dent*. 2011;71(SUPPL. 1):S20-S33. doi:10.1111/J.1752-7325.2011.00223.X

172. McBride CM, Koehly LM, Sanderson SC, Kaphingst KA. The behavioral response to personalized genetic information: will genetic risk profiles motivate individuals and families to choose more healthful behaviors? *Annu Rev Public Health*. 2010;31:89-103. doi:10.1146/ANNUREV.PUBLHEALTH.012809.103532

173. Jacobs C, Turbitt E, McEwen A, Atkins L. Australasian Genetic Counselors' Perceptions of Their Role in Supporting Clients' Behavior Change. *J Pers Med*. 2022;13(1). doi:10.3390/JPM13010030

174. Rizany I, Hariyati RTS, Handayani H. Factors that affect the development of nurses' competencies: a systematic review. *Enfermería Clínica*. 2018;28:154-157. doi:10.1016/S1130-8621(18)30057-3

175. Zureigat B, Gould D, Seven M. Educational Interventions to Improve Nurses' Competency in Genetics and Genomics: A Scoping Review. *J Contin Educ Nurs*. 2022;53(1):13-20. doi:10.3928/00220124-20211210-06

176. Sharp L, Rannus K, Nurse H, Olofsson A, Kelly D, et al. Patient safety culture among European cancer nurses-An exploratory, cross-sectional survey comparing data from Estonia, Germany, Netherlands, and United Kingdom. *J Adv Nurs*. 2019;75:3535-3543. doi:10.1111/jan.14177

177. Albada A, Vernooij M, Osch L van, Pijpe A, Dulmen S van, Ausems MGEM. Does and should breast cancer genetic counselling include lifestyle advice? *Fam Cancer*. 2014;13(1):35-44. doi:10.1007/S10689-013-9672-5

178. Han CJ, Lee YJ, Demiris G. Interventions Using Social Media for Cancer Prevention and Management: A Systematic Review. *Cancer Nurs*. 2018;41(6):E19-E31. doi:10.1097/NCC.0000000000000534

179. Valero-Aguilera B, Bermúdez-Tamayo C, García-Gutiérrez JF, Jiménez-Pernett J, Vázquez-Alonso F, Suárez-Charneco A, et al. Factors related to use of the Internet as a source of health information by urological cancer patients. *Support Care Cancer*. 2012;20(12):3087-3094. doi:10.1007/s00520-012-1431-x

180. Allen CG, Roberts M, Andersen B, Khoury MJ. Communication About Hereditary Cancers on Social Media: A Content Analysis of Tweets About Hereditary Breast and Ovarian Cancer and Lynch Syndrome. *J Cancer Educ*. 2020;35(1):131-137. doi:10.1007/S13187-018-1451-4

181. Chretien KC, Kind T. Social media and clinical care: Ethical, professional, and social implications. *Circulation*. 2013;127(13):1413-1421. doi:10.1161/CIRCULATIONAHA.112.128017/-/DC1

182. Diez de los Rios de la Serna C, Fernández-Ortega P, Lluch-Canut T. Educational Programme for Cancer Nurses in Genetics, Health Behaviors and Cancer Prevention: A Multidisciplinary Consensus Study. *J Pers Med*. 2022;12(7):1104. doi:10.3390/jpm12071104

138. Kerr D, Ostaszkiewicz J, Dunning T, Martin P. The effectiveness of training interventions on nurses' communication skills: A systematic review. *Nurse Educ Today*. 2020;89:104405. doi:10.1016/J.NEDT.2020.104405

184. Young AL, Butow PN, Tucker KM, Wakefield CE, Healey E, Williams R. When to break the news and whose responsibility is it? A cross-sectional qualitative study of health professionals' views regarding disclosure of BRCA genetic cancer risk. *BMJ Open*. 2020;10(2):e033127. doi:10.1136/BMJOPEN-2019-033127

185. Katapodi MC, Jung M, Schafenacker AM, Milliron KJ, Mendelsohn-Victor KE, Merajver SD, et al. Development of a web-based family intervention for BRCA carriers and their biological relatives: Acceptability, feasibility, and usability study. *J Med Internet Res*. 2018;20(4):1-13. doi:10.2196/cancer.9210

186. Hegarty J, Egan SM, Jones MM, Odisigo C, O'flaherty SJ, Chakraborty S, et al. *The Unmet Need in Cancer Genetic Services: Conducting an Environmental Scan of the Cancer Genetics Services in an Irish Context Underpinned by a Mixed Methods Approach*.; Report prepared for the Irish Cancer Society, Ireland.2021. Accessed November 2, 2023. https://www.cancer.ie/sites/default/files/2021-04/Conducting an environmental scan of the cancer genetics services Report 2021.pdf

187. Saifan A, Devadas B, Daradkeh F, Abdel-Fattah H, Aljabery M, Michael LM. Solutions to bridge the theory-practice gap in nursing education in the UAE: a qualitative study. *BMC Med Educ*. 2021;21(1):1-11. doi:10.1186/S12909-021-02919-X/TABLES/3

188. Calzone KA, Kirk M, Tonkin E, Badzek L, Benjamin C, Middleton A. Increasing nursing capacity in genomics: Overview of existing global genomics resources. *Nurse Educ Today*. 2018;69:53-59. doi:10.1016/J.NEDT.2018.06.032

189. Peppercorn J. Financial Toxicity and Societal Costs of Cancer Care: Distinct Problems Require Distinct Solutions. *Oncologist*. 2017;22(2):123. doi:10.1634/THEONCOLOGIST.2016-0301

190. Pichler T, Mumm F, Dehar N, Dickman E, Díez de Los Ríos de la Serna C, Dinkel A, et al. Understanding communication between patients and healthcare professionals regarding comprehensive biomarker testing in precision oncology: A scoping review. *Cancer Med*. 2024;13(3):1-15. doi:10.1002/cam4.6913

191. Fernández-Ortega P, Dies de los Ríos de la Serna C. Cancer nurses, are we really contributing to reduce burden via cancer prevention? *Asia-Pacific J Oncol Nurs*. 2022;9(6):100066. doi:10.1016/j.apjon.2022.04.005

192. Diez de los Ríos de la Serna C, Fernández-Ortega P. Enfermeros en la prevención del cáncer: cómo llegar a los que no se llega. *Magna Sci UCEVA*. 2022;2(1):75-81. doi:10.54502/msuceva.v2n1a8

193. Diez de los Rios de la Serna C, Fernandez-Ortega P, Lluch-Canut T. CN13 Review of healthcare interventions to promote cancer prevention by improving lifestyle behaviours. *Ann Oncol*. 2021;32:S1260. doi:10.1016/J.ANNONC.2021.08.638

194. Abstracts from the 55th European Society of Human Genetics (ESHG) Conference: e-Posters. *Eur J Hum Genet 2023 311*. 2023;31(1):91-344. doi:10.1038/s41431-023-01339-3

Annexes

Annex 1: Ethics committee approval

Annex 2: Participant information and consent form

Annex 3. Letter of Acceptance of Seminars in Oncology Nursing

Annex 4: Letters of permission for European Mention

Annex 1: Ethics committee approval

Comissió de Bioètica
Universitat de Barcelona

Dictamen favorable – Enmienda N°003-2022

La Comissió de Bioètica de la Universitat de Barcelona (CBUB), en sessió ordinària el dia 14 de juny de 2021, va emitir informe favorable, un cop avaluats els aspects metodològics, ètics i legals de la tesi doctoral "Nurse's role in the promotion of lifestyle behaviours in a population with genetic predisposition to cancer" a càrrec de la doctoranda Celia Diez de los Ríos de la Serna, dirigida per la Dra. María Teresa Lluch Canut.

El 30 de desembre de 2021, la doctoranda Diez de los Ríos de la Serna va presentar una esmena al projecte. La Secretaria de la CBUB va sol·licitar major informació, que va ser avaluada favorablement el 10 de novembre de 2022.

En Barcelona, a 10 de novembre de 2022.

Firmado por JORDI GARCIA FERNANDEZ - DNI ***3005** (TCAT) el día 20/02/2023 con un certificado emitido por EC-SectorPublic

Dr. Jordi García Fernández
Vicerector de Recerca
President de la Comissió de Bioètica
Universitat de Barcelona

Institutional Review Board (IRB00003099)

Annex 2: Participant information and consent form

Participant Information Sheet

(Delivered in each phase of the study)

1. **Study title**

Nurses' role in the promotion of lifestyle behaviours in a population with genetic predisposition to cancer

2. **Invitation paragraph**

You are being invited to take part in a research study. Before you decide, it is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully and discuss it with others if you wish. Ask us if there is anything that is not clear or if you would like more information.

3. **What is the purpose of the study?**

This study is carried out in the University of Barcelona and its aim is to develop and evaluate a nurse-led educational programme to promote a healthy lifestyle in individuals with high susceptibility to cancer.

1. Phase 1: The study will take place in several countries across Europe. Our project includes many stages and activities. In this first task we want to explore what nursing interventions are used and needed during genetic counselling to promote a healthy lifestyle in people with high-risk cancer and select what is necessary and what healthcare professionals consider to be of utmost importance in the context of promoting healthy lifestyle behaviours for patients with risk of cancer.

   Our project will involve online Delphi surveys. **We are looking to recruit up to 36 healthcare professionals with experience looking after/treating patients with high risk of cancer.** When we have enough people taking part in this study, we will not include or invite any more people.

2. Phase 2: The study will take place in several countries across Europe. Our project includes many stages and activities. In this task we want to explore the experience and the key points patients feel healthcare professionals should learn in order to support their experience.

   Our project will involve semi-structured interviews. **We are looking to recruit up to 15 people with genetic predisposition to breast cancer and colorectal cancer.** When we have enough people taking part in this study, we will not include or invite any more people.

This research will allow us to identify key areas that require special attention and intervention to improve the promotion of healthy lifestyle habits and support people at risk of cancer.

4. Why have I been invited to participate?

1. Phase 1 You have been invited to take part in this study because you are a **healthcare professional with experience looking after/treating patients with cancer or individuals with high risk of cancer** (oncologists, oncology nurses, genetic counsellor, nutritionist, physical therapist, psychologist or other professionals involved in the treatment of these patients).

2. Phase 2 You have been invited to take part in this study because you are a **person who has been identified with high risk of breast cancer or colorectal.**
   You can only be part of this study if you are older than 18 years old.

3. Do I have to take part? What will happen to me if I take part?

No. It is up to you to decide whether to take part.

If you decide to take part, you are still free to withdraw at any time and without giving a reason. If you decide to withdraw, we will delete any personal data we collect as part of the study that could identify you.

If you require any additional information, you will be able to contact the researchers by telephone or email at this stage.

To participate in the online Delphi survey:

To participate, you will be required to take the survey for different rounds. There will be questions to be answered in each round that will take approximately 15-20 minutes to complete.

To participate in the interviews:

To participate, you will be invited to an online interview that will take approximately 40 minutes.

4. What are the possible disadvantages and risks of taking part?

As this study does not directly affect you, there are no real disadvantages of taking part. However, some people might find taking part in surveys/interviews time-consuming. If you did feel that the survey/interview was taking longer than you thought, you can stop, save your progress, and return to it later. Or you can completely withdraw from the study without your decision having any effect whatsoever.

By taking part in the interview, research participants (people with high risk of breast cancer or colorectal cancer) might find thinking on their experience upsetting and feel a level of

discomfort. We advise participants who are upset that we can stop the interview at any time or delay.

5. What will happen to my data?

This study has been approved by the Research Ethics Committee of the University of Barcelona.
According to the General Data Protection **Regulation** (**EU**) **2016/679** (GDPR) on data protection and privacy and Organic Law 3/2018, of December 5, all personal information will be kept strictly confidential. Your name or identifiable information will not appear in any publication. The data will be identified by a code and only the collaborators in the study will have access to it. The study files will be stored on a secure server such as the UB-cloud under a password only known to the researcher.

In accordance with the provisions of the aforementioned regulation, the UNIVERSITY OF BARCELONA, (with CIF Q0818001J and domicile in Gran Via de les Corts Catalanes, 585 -08007) as responsible for the processing of personal data, informs you that you can contact the Data Protection Officer by writing to the postal address (Travessera de les Corts, 131-159, Pavelló Rosa, 08028 - Barcelona), or by email to protecciodedades@ub.edu

You have the right to access your data, to request the rectification of inaccurate data and to request its deletion, as well as to limit the processing, to object and to withdraw consent to its use for certain purposes. You can do this by writing to the postal address or by e-mail to the address mentioned in the preceding paragraph. Likewise, we inform you of your right to file a complaint with the Catalan Data Protection Agency in the case of any action of the University of Barcelona that you consider to violate your rights.

6. Contact for Further Information

PhD student, Celia Díez de los Ríos. Escuela de enfermería. UB.

Email: cdiezdde7@alumnes.ub.edu

Thank you for your time, interest and collaboration.

Title of Project: Nurses' role in the promotion of lifestyle behaviours in a population with genetic predisposition to cancer

Name of Researcher(s): PhD Student Celia Díez de los Ríos

CONSENT FORM

1. I confirm that I have read and understood all the all the information about the project.	YES/NO
2. I have had the opportunity to think about the information and ask questions, and I understand the answers I have been given.	YES/NO
3. I have been informed about the project by the investigator.	YES/NO
4. I have received enough information about the project.	YES/NO
5. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason and without being affected in any way.	YES/NO
6. I have been told and I understand the possible risks of my participation in the study.	YES/NO
7. I agree to the way my data will be collected and processed, and that research data will be stored by the investigators in accordance with relevant Data Protection policies and regulations (*Ley Orgánica 3/2018, de 5 de diciembre, de Protección de Datos Personales y garantía de los derechos digitales*).	YES/NO
8. I understand that all personal and research data and identifiable information I provide will be kept confidential and will be seen only by the investigators.	YES/NO
9. I understand that information I provide during my survey may be quoted in reports and articles that are published about the study, but my name or anything else that could identify me will not be revealed.	YES/NO
10. I agree to take part in the study.	YES/NO

_____	_____	_____
Name of participant	*Date*	*Signature*
_____	_____	_____
Investigator	*Date*	*Signature*

(1 copy for participant; 1 copy for investigator)

In case you want to ask any further questions about the project or if you want to withdraw consent to participate in the study please contact: Celia Díez de los Ríos. Escuela de enfermería. UB.

Email: cdiezdde7@alumnes.ub.edu

Hoja de información y consentimiento informado

(Se usará en cada fase del estudio)

1. **Título del estudio:**

Rol de las enfermeras en la promoción de estilos de vida en personas con predisposición genética al cáncer

Nos dirigimos a usted para informarle acerca de un estudio de investigación que se está llevando a cabo en nuestro centro y en el que se le invita a participar. Es importante que usted reciba la información necesaria para poder decidir si quiere o no participar en este estudio.Para ello lea con detenimiento este documento y nosotros le aclararemos las dudas que le puedan surgir. Su participación es voluntaria y puede decidir no participar o cambiar su decisión y retirar su consentimiento en cualquier momento sin que esto repercuta en sus cuidados.

2. **¿Cuál es el propósito del estudio?**

Este estudio se realiza en la Universidad de Barcelona y su objetivo es desarrollar y evaluar un programa educativo dirigido por enfermeras para promover un estilo de vida saludable en personas con alta susceptibilidad al cáncer.

1. Fase 1 El estudio se llevará a cabo en varios países de Europa. Nuestro proyecto incluye muchas etapas y actividades. En esta primera etapa queremos explorar las intervenciones de enfermería que se realizan y necesitan durante el asesoramiento genético para promover un estilo de vida saludable en personas con cáncer de alto riesgo y seleccionar lo que es necesario y lo que los profesionales de la salud consideran de suma importancia en el contexto de la promoción de comportamientos de estilo de vida saludable para pacientes con riesgo de cáncer.

   Nuestro proyecto incluirá encuestas Delphi. **Buscamos reclutar hasta 36** profesionales **de la salud con experiencia en el cuidado/tratamiento de pacientes con alto riesgo de cáncer.** Cuando tengamos suficientes personas participando en este estudio, no incluiremos ni invitaremos a más personas.

1. Fase 2: El estudio se llevará a cabo en varios países de Europa. Nuestro proyecto incluye muchas etapas y actividades. En esta tarea queremos explorar la experiencia y los puntos clave que los pacientes sienten que los profesionales de la salud deben aprender para apoyar su experiencia.

   Nuestro proyecto incluirá entrevistas semiestructuradas. **Buscamos reclutar hasta 15 personas con predisposición genética al cáncer de mama y cáncer colorrectal.** Cuando tengamos suficientes personas participando en este estudio, no incluiremos ni invitaremos a más personas.

   Esta investigación nos permitirá identificar áreas clave que requieren especial atención e intervención para mejorar la promoción de hábitos de vida saludables y apoyar a las personas en riesgo de cáncer.

3. ¿Por qué me han invitado a participar?

1. Fase 1 Se le ha invitado a participar en este estudio porque usted es un profesional de la **salud con experiencia en el cuidado /tratamiento de pacientes con cáncer o personas con alto riesgo de cáncer** (Oncólogos, enfermero oncológico, asesor genético, nutricionista, fisioterapeuta...)

2. Fase 2 Se le ha invitado a participar en este estudio porque es una **persona con alto riesgo de cáncer de mama o cáncer colorrectal.**
   Solo puede ser parte de este estudio si es mayor de 18 años.

4. ¿Tengo que participar? ¿Qué me pasará si participo?

No. Puedes decidir libremente si participas.

Si usted decide participar, todavía es libre de retirarse en cualquier momento y sin necesidad de dar ninguna razón. Si decide retirarse, destruiremos cualquier dato personal que recopilemos como parte del estudio que pueda identificarle.

Si necesita alguna información adicional, podrá ponerse en contacto con los investigadores por correo electrónico.

Para participar en la encuesta en línea de Delphi:

Para participar, deberá realizar la encuesta durante varias rondas. Habrá 10-15 preguntas en cada ronda que tardarán aproximadamente 15-20 minutos en completarse.

Para participar en las entrevistas:

Para participar, se le invitará a una entrevista online que durará aproximadamente 40 minutos.

5. ¿Cuáles son las posibles desventajas y riesgos de participar?

Como este estudio no le afecta directamente, no hay desventajas reales de participar. Sin embargo, algunas personas pueden sentir que participar en encuestas consume mucho tiempo. Si es así las encuestas pueden retomarse, guardar su progreso y volver a ella más tarde. O puede retirarse completamente del estudio sin problema.

Al participar en la entrevista, los participantes de la investigación (personas con alto riesgo de cáncer de mama o cáncer colorrectal) pueden encontrar molesto pensar en su experiencia y sentirse incómodos. Informamos a los participantes que se sientan así que podemos detener la entrevista en cualquier momento o retrasarla.

6. Confidencialidad ¿Qué pasará con mis datos?

Este estudio ha sido aprobado por el Comité de Ética investigadora de la Universitat de Barcelona.

En cumplimiento del Reglamento 2016/679 de la Unión Europea, de 27 de abril, relativo a la protección de las personas físicas en lo que respecta al tratamiento de sus datos personales y a la Ley Orgánica 3/2018, de 5 de diciembre, toda la información sobre usted, o las respuestas que proporcione, durante el transcurso de este estudio se mantendrá estrictamente confidencial. Su nombre o información identificable no aparecerá en ninguna publicación. Los

datos serán identificados por un código y sólo los colaboradores en el estudio tendrán acceso a los mismo. los archivos de estudio se almacenarán en un servidor seguro como la UB-cloud bajo una contraseña conocida solo por el investigador.

De conformidad con lo establecido en la mencionada regulación, la UNIVERSIDAD DE BARCELONA, (con CIF Q0818001J y domicilio en la Gran Via de les Corts Catalanes, 585 -08007 Barcelona) como responsable del tratamiento de los datos personales, le informa que puede contactar con el Delegado de Protección de Datos mediante escrito a la dirección postal (Travessera de les Corts, 131-159, Pavelló Rosa, 08028 - Barcelona),o mediante un mensaje de correo electrónico a protecciodedades@ub.edu

Usted tiene derecho a acceder a sus datos, a solicitar la rectificación de los datos inexactos y a solicitar su supresión, así como a limitar el tratamiento, a oponerse y a retirar el consentimiento de su uso para determinadas finalidades. Estos derechos los puede ejercer mediante escrito a la dirección postal o mediante un mensaje de correo electrónico a la dirección mencionada en el párrafo anterior. Así mismo, le informamos de su derecho a presentar una reclamación ante la Agencia Catalana de Protección de Datos en el caso de cualquier actuación de la Universitat de Barcelona que considereque vulnera sus derechos.

7. Para más información contacte con:

Celia Díez de los Ríos. Escuela de enfermería. UB.

Email: cdiezdde7@alumnes.ub.edu

Gracias por leer la Hoja de Información

Título del proyecto: Rol de las enfermeras en la promoción de estilos de vida en personas con predisposición genética al cáncer

Investigadora: Celia Díez de los Ríos (Estudiante de Doctorado)

Hoja de consentimiento

1. Confirmo que he leído y entendido toda la información sobre el proyecto	SI/NO
2. He tenido la oportunidad de pensar en la información y hacer preguntas, y entiendo las respuestas que se me han dado.	SI/NO
3. He sido informado sobre el proyecto por el investigador	SI/NO
4. He recibido suficiente información sobre el proyecto	SI/NO
5. Entiendo que mi participación es voluntaria y que soy libre de retirarme en cualquier momento, sin dar ninguna razón y sin ningún perjuicio	SI/NO
6. Me han informado y entiendo los posibles riesgos de mi participación en el estudio	SI/NO
7. Acepto la forma en que mis datos serán recogidos y procesados, y que los datos de investigación se almacenarán en las instalaciones de archivo de la Universidad de acuerdo con las políticas y regulaciones pertinentes de protección de datos. (*Ley Orgánica 3/2018, de 5 de diciembre, de Protección de Datos Personales y garantía de los derechos digitales*).	SI/NO
8. Entiendo que todos los datos personales y de investigación y toda información identificable que proporcione, se mantendrán confidenciales y sólo serán vistos por investigadores de la Universidad de Barcelona.	SI/NO
9. Entiendo que la información que proporcione durante la investigación puede ser citada en informes y artículos que se publican sobre el estudio, pero mi nombre o cualquier otra cosa que pueda identificarme no será revelada.	SI/NO
10. Estoy de acuerdo en participar en el estudio.	SI/NO

_____	_____	_____
Nombre del participante	*Fecha*	*Firma*
_____	_____	_____
Investigador	*Fecha*	*Firma*

(1 copia para participante; 1 copia para investigador)

En caso de que desee hacer más preguntas sobre el proyecto o si desea retirar el consentimiento para participar en el estudio, póngase en contacto con: Celia Díez de los Ríos. Escuela de enfermería. UB. **Email**: cdiezdde7@alumnes.ub.edu

Annex 3: Letter of Acceptance of Seminars in Oncology Nursing

Date:	Mar 01, 2024
To:	"Celia Diez de los Rios de la Serna" celia.diezdelosriosdelaserna@glasgow.ac.uk;research.assistant@cancernurse.eu
From:	"Maura Dowling" maura.dowling@universityofgalway.ie
Subject:	Decision on submission to Seminars in Oncology Nursing

Manuscript Number: **SONU-D-23-00278R2**

Hereditary cancer syndrome carriers: feeling left in the corner

Dear Celia

Thank you for submitting the final minor edits to your manuscript to Seminars in Oncology Nursing.

I am pleased to inform you that your manuscript has been accepted for publication.

Your accepted manuscript will now be transferred to our production department. We will create a proof which you will be asked to check, and you will also be asked to complete a number of online forms required for publication. If we need additional information from you during the production process, we will contact you directly.

We appreciate you submitting your manuscript to Seminars in Oncology Nursing and hope you will consider us again for future submissions.
We encourage authors of original research papers to share the research objects – including raw data, methods, protocols, software, hardware and other outputs – associated with their paper. More information on how our open access Research Elements journals can help you do this is available at https://www.elsevier.com/authors/tools-and-resources/research-elements-journals?dgcid=ec_em_research_elements_email.

Kind regards,

Maura
Maura Dowling, PhD, MSc, BNS, RNT, RGN
Associate Editor
Seminars in Oncology Nursing

Annex 4: Letter from the Doctoral School approving the application for the international

UNIVERSITAT DE BARCELONA

Escola de Doctorat

Notificació d'acords presos per la Comissió Acadèmica del programa de doctorat

Sra. Celia Díez de los Ríos de la Serna

D'acord amb el que disposa l'article 40 de la llei 39/2015, d'1 d'octubre , del Procediment Administratiu Comú de les Administracions Públiques, per la present us notifico que en data 3 de juliol de 2023, per la Comissió Acadèmica del programa de doctorat de Infermeria i Salut de la Universitat de Barcelona, ha estat dictat el següent acord.

S'autoritza l'estada de recerca sol·licitada per la Sra. Celia Díez de los Ríos de la Serna al centre: Escola d'Infermeria de la Universidad de Galway entre agosto y octubre de 2023 Durant el següent període:
D'agost fins a l'octubre de 2023

Barcelona, a 4 de juliol de 2023

(signatura)

NÚRIA FABRELLAS PADRÉS - DNI 77999391C

Nuria Fabrellas

Presidenta de la Comissió Acadèmica

Contra els acords presos per la Comissió Acadèmica referents a sol·licitud d'acceptació del pla de recerca establert en l'article 33.7 i contra l'avaluació anual del pla de recerca establert en l'article 34, es pot interposar recurs d'alçada davant del rector de la Universitat de Barcelona, d'acord amb el que disposa l'article 79 dels Estatuts de la Universitat de Barcelona. El recurs es pot interposar en el termini d'un mes, a comptar de l'endemà de la notificació segons s'estableix en els articles 121 i 122 de la Llei 39/2015, d'1 d'octubre, del Procediment Administratiu Comú de les Administracions Públiques.

“Valeu a pena? Todo vale a pena

Se a alma não é pequena”

Mensagem

Fernando Pessoa