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Oncogenetic counselling in Portugal: A literature review

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Dedicatória

Para a minha família que sempre acreditou em mim, me apoiou e me guiou durante todo o meu percurso académico.

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Resumo

Introdução: Aconselhamento genético é uma área cada vez mais presente, especialmente o ramo de oncogenética devido à prevalência das síndromes neoplásicos hereditários. Dado o desenvolvimento de técnicas de identificação e diagnóstico de variantes patogénicas, levou a que mais utentes e as suas famílias tivessem ao seu dispor testes genéticos. Neste sentido, a introdução de aconselhamento genético (AG) nos serviços de saúde tornou-se imprescindível para que os utentes e suas famílias que passam por este processo possam ter algum acompanhamento especializado que os ajude a nível psicológico, social, pessoal e profissional durante todo o processo. Em Portugal, a especialidade de Genética Médica foi reconhecida pela Ordem dos Médicos em 1998, disponibilizando desde essa data a consulta de AG a todos os utentes, não obstante, esta continua a ser uma área com pouca visibilidade e conhecimento por parte da população médica, assim como da população em geral.

Objetivos: O objetivo principal desta revisão é perceber a organização dos serviços de AG na área oncológica em Portugal, quais as organizações e recomendações existentes. Compreender a envolvência desta temática no SNS, e a sua participação a nível europeu.

Métodos: Pesquisa bibliográfica na base de dados *Pubmed, ResearchOnly, UptoDate e Google Scholar*. Uso de referências bibliográficas de artigos selecionados, como também o uso de livros e jornais indexados, relacionados com a temática da dissertação.

Resultados: Portugal encontra-se numa fase de desenvolvimento, com legislação publicada referente a testes genéticos e AG. Iniciou também um mestrado profissionalizante nesta área, de maneira a aumentar a formação que é escassa. Portugal faz parte de associações europeias que visam desenvolver diretrizes consensuais a nível internacional, como também tem uma associação de profissionais de AG a nível nacional (APPAcGen). Esta associação engloba todos os aspetos de AG, como a organização dos serviços no SNS, critérios de referência dos utentes, entre outras informações relevantes tanto para profissionais de saúde como para a população em geral. Na área de oncogenética, o facto de no SNS haver hospitais oncológicos levou a que esta área fosse desenvolvida e fosse criado guias de orientação terapêutica para estes pacientes e suas famílias. Estes guias ajudam os profissionais de saúde a referenciar os pacientes quando necessário, como também a identificar a necessidade de AG para estes casos. Existe neste momentos alguns serviços com AG integrado, porém continua a ser escasso e pouco suficiente para toda a população que necessita destes cuidados.

Conclusão: Em Portugal, diferente do que acontece em outros países europeus, o AG é da responsabilidade do médico geneticista e não de um técnico de AG. Há um grande desconhecimento no que concerne a esta área quer pela população em geral, quer pelos

próprios profissionais de saúde, como uma necessidade cada vez maior de ter equipas multidisciplinares nesta área

Palavras-chave

Educação genética; aconselhamento genético; serviços portugueses de genética oncológica; Oncogenética; aconselhamento oncogenético

Resumo Alargado

Introdução: O aconselhamento genético (AG) tem-se tornado uma área cada vez mais presente na profissão médica devido a toda a evolução científica e tecnológica associada ao conhecimento e diagnóstico de doenças genéticas. O facto de cada vez mais existirem novos métodos científicos para identificação de variantes patogénicos, levou a que fosse necessário a introdução de aconselhamento genético nesta área, de maneira a ajudar os pacientes e suas famílias a lidarem com este tipo de situações a nível social, pessoal, profissional e psicológico. Sendo as síndromes neoplásicas hereditárias parte deste grupo, como por exemplo o Síndrome do cancro hereditário da mama e do ovário, e sendo também condições com bastante investigação e reconhecimento associado, o aconselhamento oncogenético tornou-se um ramo do AG, para o qual é necessário especial atenção. Os técnicos de AG, na área da oncogenética necessitam de um conjunto de competências que os permitam realizar uma investigação de padrões de hereditariedade e de risco em famílias que seguem hereditariedade mendeliana, como também acompanhar e dar apoio psicossocial durante todo o processo envolvente. Necessitam também de ter conhecimento das estruturas aptas dentro do sistema nacional de saúde de maneira a conseguir a correta referenciação destes pacientes, como também saber quais as opções de tratamento e acompanhamento disponíveis para estes casos. Em 1998 a especialidade de Genética Médica foi reconhecida pela Ordem dos Médicos e tornou-se parte integrante do Sistema Nacional de Saúde (SNS). Este facto aumenta a necessidade de integração profissional do aconselhamento genético nos serviços de genética médica do SNS, que atualmente ainda é escassa devido à falta de profissionais especializados na área. Em Portugal, o AG é realizado por oncologistas e geneticistas.

Objetivos: Desta forma, o objetivo desta revisão de literatura é perceber o nível dos serviços de aconselhamento genético na área oncológica em Portugal, compreender a envolvência desta temática no SNS, e a sua participação a nível europeu. Como objetivos secundários, esta revisão de literatura visa identificar a evolução da prática de AG em Portugal, descobrir novas oportunidades de desenvolvimento, de melhoria e comparar práticas nacionais com práticas de outros países europeus através de uma revisão descritiva da literatura.

Métodos: Para esta dissertação, foi realizado uma pesquisa bibliográfica na base de dados *Pubmed, ResearchOnly, Google Scholar e UpToDate,* utilizando as palavras-chave referentes a esta dissertação, tanto em Inglês como em Português: Educação genética; aconselhamento genético; serviços portugueses de genética oncológica; Oncogenética; aconselhamento oncogenético. A investigação e o desenvolvimento desta área de estudo tem tido como principais figuras Milena Paneque, Jorge Sequeiros e Heather Skirton. Pela sua dedicação e vasta contribuição, a presente revisão focou-se, portanto, nos referidos autores. Foi utilizado também livros e jornais indexados relacionados com a temática da dissertação.

Resultados: De acordo com a bibliografia pesquisada, é de notar que Portugal encontra-se numa fase de desenvolvimento e evolução, porém com algumas limitações em seus serviços de genética médica, especialmente pela falta de técnicos de AG. O facto de existir um mestrado profissionalizante em AG implica que a necessidade destes serviços tem aumentado, e que a formação específica tem sido necessária. A implementação de legislações relacionadas dirigidos as AG e testes genéticos, demonstra também a evolução de Portugal, e o início do reconhecimento desta profissão. Como parte integrante de organizações internacionais como a European Society of Human Genetics, Portugal estabelece-se como um pais europeu que está a tentar desenvolver esta área no SNS, porém ainda não existe reconhecimento dos técnicos de AG a nível nacional, o que dificulta a sua integração. Não obstante este facto, Portugal tem a Associação Portuguesa de Profissionais de Aconselhamento Genético (APPAcGen), que visa aumentar a conscientização da população, de maneira a conseguir o reconhecimento necessário. Porém, existem ainda várias limitações, como a falta de profissionais especializados, integração dos mesmos em equipas multidisciplinares e o pouco conhecimento das redes nacionais de genética médica e dos serviços disponíveis a nível do SNS. Porém, na área de Oncogenética e através do Instituto Português de Oncologia (IPO), já foi desenvolvido um guia de orientação terapêutica para utentes que necessitem de acompanhamento genético, a nível de testes como também de aconselhamento. Atualmente existem alguns serviços que já integram o AG, porém continua a ser escasso e pouco suficiente para toda a população que necessita destes cuidados.

Conclusão: Portugal demonstra estar a evoluir nesta temática, porém necessita da integração de técnicos de AG no SNS, como também do reconhecimento dos mesmos para que esta integração seja possível. A nível das síndromes hereditárias neoplásicas, Portugal aparenta ter iniciado o processo de integração destes serviços ao ter disponíveis guias de orientação para profissionais de saúde, como também legislação referente a este tema. Comparativamente com outros países europeus, Portugal necessita de desenvolver a sua formação especializada, tanto a nível graduado como a profissionais de saúde que lidam com estes casos, para que hajam mais recursos disponíveis para os utentes e suas famílias. O AG em Portugal é realizado pelos médicos, que ao requisitarem testes genéticos têm que fazer o adequado AG a cada paciente. O reconhecimento de técnicos de AG poderá ser uma das opções futuras para o nosso país, como já acontece em outros países europeus.

Palavras-chave

Educação genética; aconselhamento genético; serviços portugueses de genética oncológica; Oncogenética; aconselhamento oncogenético

Abstract

Introduction: Genetic counselling is an increasingly current subject area especially its branch of oncogenetics due to the prevalence of hereditary neoplastic syndromes. The scientific and technological evolution associated with this matter has made genetic testing available to the population, increasing the need and importance of introducing genetic counselling into the national health system (NHS) for appropriate guidance and support of patients and their families. However, this is still an area with little visibility and awareness by other medical professionals, which translates as few specialised professionals, and therefore, scarce specialised services. This portrays that the awareness for the inclusion of these services within the national health system (NHS) is present, however, there are still barriers that need to be overcome to fully reach all the patients and families in need.

Objective: The main objective of this study is to understand how Portugal has developed its genetic counselling practice and how it is organised at a national level. As secondary objectives this review aims to Identify development of genetic counselling in Portugal, and how it has been able to reach all the patients and families in need of these services, identify new opportunities for development and enhancement of genetic counselling practice in Portugal and compare genetic counselling practice in Portugal with other European countries, through a descriptive literature review.

Methods: Research of scientific articles in the *Pubmed, ResearchOnly, Google Scholar and UpToDate* databases, and use of bibliographic references.

Results: Portugal has initiated this introduction by creating a master's degree, having legislation related to the subject, having developed guidelines and recommendation for oncogenetic patients, and by having a national association of genetic counsellors (APPAcGen), as well as be part of international associations, such as the European Society of Human Genetics. Genetic counselling is integrated into some medical services and is part of the medical practice in Portugal, however, there is still a lack of specialised professionals and integration of these in multidisciplinary teams within the NHS.

Conclusion: Portugal has initiated the development of this area within its services, however, as it still is an unrecognised medical profession, its inclusion within the NHS is hard and has shown to have many barriers. At a European level, Portugal is heading the right direction, accompanying many other European countries in the development of this subject area.

Keywords

Genetic education; Genetic Counselling; Portuguese cancer genetics services; Oncogenetic; Cancer genetic counselling

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AG	Aconselhamento Genético
APC	Adenomatous Polyposis Coli
APPAcGen	Associação Portuguesa de Profissionais de Aconselhamento Genético
BRCA1	Breast Cancer 1
BRCA2	Breast Cancer 2
CGC	Centro de Genética Clinica
CGPP	Centro de Genética Preditiva e Preventiva
CNECV	Conselho Nacional de Ética para as Ciências da Vida
DGS	Direção Geral de Saúde
EBMG	European Board of Medical Genetics
EPCAM	Epithelial Cell Adhesion Molecule
ESHG	European Society of Human Genetics
FAP	Familial Adenomatous Polyposis
GNGC	Genetic Nurses and Genetic Counsellors
HBOC	Hereditary Breast and Ovarian Cancer
IBMC	Instituto de Biologia Molecular e Celular
ICBAS	Instituto de Ciências Biomédicas Abel Salazar
INSA	Instituto Nacional de Saúde Dr. Ricardo Jorge
IPO	Instituto Português de Oncologia
LS	Lynch Syndrome
MD	Medical Degree
MLH1	MutL Homolog 1
MMR	MisMatch Repair
MS	Ministério da Saúde
MSc	Masters in Science
MSH2	MutS Homolog 2
MSH6	MutS Homolog 6
NHS	National Health System
OM	Ordem dos Médicos
PMS2	PMS1 Homolog 2
PST	Pre Symptomatic Testing
SNS	Sistema Nacional de Saúde
UK	United Kingdom
UP	Universidade do Porto
USA	United States of America
WHO	World Health Organisation

Introduction

Medical genetics has been a developing area in the medical field since de the 1990s. When in 2003, through the Human Genome Project, the mapping sequence of the human genome was completed, the medical field was introduced to a *new era* of genomics, identified as*: "the study of the entire human genome; as applied to health outcomes, genomic research includes interactions of multiple genes with each other and with the environment."* Since then, it was possible for the investigation of multiple genes and their interaction with health outcomes and influence on diseases, such as cancer. (1)

The World Health Organisation (WHO) declares genetic counselling as a field for the understanding of medical genetics to the general population, as well as, allowing prevention and identification of genotypes responsible for hereditary conditions by means of a retrospective or prospective investigation of genetic material. WHO has also stated that genetic counselling should be available to patients and their families, in both developed and developing countries. (2)

This *new era* also introduced genomic testing, which allowed the testing and identification of single genes and pathogenic variants, which are genetic alteration (mutation) that increases an individuals' susceptibility of risk to a certain condition. This genetic testing is carried out by patients with the condition, and if a familial mutation is identified, then presymptomatic testing (PST) should be carried out in asymptomatic relatives. Genetic testing also aids the investigation of pathological outcomes in these conditions, and are able to deduce risk. Given this new flood of information and the development of technological knowledge in this area, the introduction of genetic counselling is extremely necessary. (1)

As this is a recent medical area, many people are not aware of what it consists, and how important it can be for them and their families. This is where the genetic counselling comes in as an important resource of information and communication, and an important instrument to facilitate the cognitive process involved in receiving such diagnosis. It is safe to say that genetic counselling can be seen as a guidance tool to these patients and their families. (3)

The fact that the new era of genomics allows an in-depth investigation of families' susceptibility and risk probabilities of having a certain disease, emphasises the great importance to introduce genetic counselling. These families, as well as the single patients, need orientation and guidance which they see as unknown and strange. As it is a predictive territory, patients and families might not even be able to correctly interpret results, which can lead to psychological stress. (4)

As I will discuss further on, genetic counselling has had many definitions throughout the years, as well as many guidelines and recommendations, which are different in many countries. This leads to many different opinions and strategies that are developed at international level, and lack of consensus when it comes to this matter. (5)

As this is an area with a complex scientific and social interface, and also an area of study which allows the health professional to look at the patient and their families holistically, it is important to develop consensual international guidelines, which would set in motion more investigation and discussion about these issues, raising awareness within the health professionals, as well as the population in general. An example of this is the American College of Medical Genetics guidelines for referral indications for cancer predisposition assessment. (7)

Nowadays, it has been known that 5-15% of cancer cases show mendelian inheritance, that implies that these individuals are likely to carry mutations conferring high cancer risk. (1) Given this, genetic counselling of families suffering from hereditary neoplastic syndromes has been growing and becoming a crucial part of the medical field nowadays. Due to enhanced diagnostic and testing options, the need for genetic counselling services has increased exponentially, making genetic counselling crucial for the correct management of these patients and their families. For this, a partnership between specialist genetics and oncologists is essential and in some European countries, non-medical genetic counsellors are an important part of this service structure. (3)

"Cancer genetics encapsulates both genetic and oncology expertise, involving interactions between psychosocial and biomedical healthcare and the genetic technologies, and distinctive professional communities and institutionalised roles as well" (6)

There are many hereditary cancers, such as breast and ovarian cancer through the hereditary transmission of *BRAC1/BRAC2* genes, colorectal cancer in conditions such as familial adenomatous polyposis and Lynch Syndrome. Being able to apply genetic counselling to these families has been an effective way for prevention, early prognosis, early detection and also, an incentive for the correct and fully informed treatment decision. (7)

Susceptibility and risk assessment is extremely necessary in order to apply the best *modus operandi* to these families and patients, allowing the implementation of the correct preventive medicine therapy in this kind of diseases which can be named as *preventable*. As cancer has been labelled the "disease of the century" by many authors, its national economic impact is also problematic, and genetic counselling could allow the reduction of costs of treatment plans if, as above stated, the correct preventive medicine is applied to these patients. (8)

Given my background and interest in genetics, beginning prior to the initiation of my medical course, I decided to gather information on the state of the art of genetic counselling in

Portugal, focusing on the oncogenetic area, seeing as it has been a continuous area of study around the world. However, as it is a medical field in constant expansion, as a medical student, the education about this matter is scarce. As I believe this to be a very important matter, and something all health professionals should be aware, I decided this would be a great area of research for my final medical dissertation.

Objectives

The main objective of this study is to understand how Portugal has developed its genetic counselling practice, how it is organised at a national level.

The secondary objectives of this dissertation are:

1) Identify development of genetic counselling in Portugal, and how it has been able to reach all the patients and families in need of these services.

2) Identify new opportunities for development and enhancement of genetic counselling practice in Portugal.

3) Comparison of the genetic counselling practice in Portugal with other European countries, through a descriptive literature review.

Materials and Method

The initial idea for this dissertation was to make it as an investigation project, where I would interview genetic counsellors around Portugal so that I would have a more feasible set of results. However, this is not possible given the lack of participation of the professionals, due to many reasons such as time constraints, communication barriers and ethical related issues due to the privacy of patients. Hence, I had to change my strategy for this dissertation and decided on a literature review instead.

The method I used to develop this literature review was by means of research of the topic in several databases, such *as PubMed, ResearchOnly, Google Scholar* and *UpToDate*, using the keywords stated above, such as, "Portuguese cancer genetic services", "oncogenetic", "genetic counselling", "genetic education", "cancer genetic counselling" in both Portuguese and English, in order to retrieve as many articles as possible related to this subject.

As a complementary method of research, the authors that appeared to have published many articles about this matter were also researched in further depth, in order to obtain all the papers published by them. As an example, Dra Milena Paneque and Dr Jorge Sequeiros were two authors that were fully researched through the search engines, as they are both Portuguese medical professionals involved in this subject, with many papers published throughout the years. I also used google scholar to research other dissertations related to this area, as well as qualitative studies about the matter.

Using resources such as book and journals about this subject was also helpful, and I was able to use a few to develop this dissertation. The Journal of Genetic Counselling was the one that I used the most throughout my investigation, seeing as it is the one journal that is mainly focused on what I am investigating. Complimentary books about genetics, as well as genetic counselling, were used to understand terminology and also to help elaborate the educational aspect of the dissertation.

Seeing as I am focusing on the state of the art in Portugal, through search engines such as Google, I found many websites that were important to this dissertation, for example, websites of organisations and societies developed in Portugal and in other countries. Also, these websites contained important information about certain issues related to this dissertation, and that would be important to incorporate, as for example referral network layout and guidelines as well as legislation related to the matter.

Choosing the bibliography was influenced by the authors of the articles, books or websites and their impact on this field. I also took into consideration the influence of each item of

bibliography used, and their impact towards my study and how I could incorporate most of my research onto my dissertation.

Chapter 1: Contextualization of Genetic Counselling

The term genetic counselling was introduced in 1947 by Sheldon Reed as he believed it to be a new area of clinical genetics with the purpose of allowing the general population to understand the genetic problems within their families. (Álvaro Mendes 2013) Ever since the introduction of this term, this has been a developing area, and in 1975, the *Committee of Genetic Counselling of the American Society of Human Genetics* established the definition of genetic counselling that is still widely accepted.

"Genetic counselling is a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family to (1) comprehend the medical facts, including the diagnosis, probable course of the disorder, and the available management; (2) appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives; (3) understand the alternatives for dealing with the risk of recurrence; (4) choose the course of action which seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards, and to act in accordance with that decision; and (5) to make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder." (9)

It is important to emphasise that this definition has been developed in 1975, and since then there have been many changes in this field, which would lead to the readjustment of the definition of genetic counselling. Even though there have been many reviews of this definition, the one that was firstly developed is still the one that is mostly accepted within this field, and the most commonly used when it comes to defining genetic counselling. (5)

As stated by the above definition, genetic counselling aims to develop the relationship between the professional and the patient, allowing them to participate in the decision process, as well as adjusting this process in the best possible way to the individual patient and their families. It is important to understand that religion, beliefs, socio-cultural backgrounds and traditions may have a great impact on how each patient and their families adjust to diagnosis, treatment plans and also how they deal with these situations on a psychological level. Therefore, it is greatly necessary to introduce this relationship and develop a field which focuses on this matter to help and guide the patient and their families with this process. (10)

Genetic counselling allows the passing on of important individualised information, in an educational manner, enabling the patient and family to choose the best course of action,

together with the input of the professional that is helping them through this process. This manner of counselling creates a sense of responsibility and empowerment on the patient, which can allow them to be more actively involved. (11)

At the European level, there was a lack of consensus about the best way to carry out genetic counselling, which meant that there were many different approaches to this subject. Counselling has been an area that has had a widespread investigation, leading to each country coming up with its own legislations, guidelines and definitions. Due to this, at a European level, the EuroGenTest (www.eurogentest.org) was established in order to "*harmonise genetic testing across Europe.*" (12) This was a tool developed to aid professionals of genetic counselling to be able to give the best possible care to their patients and families and allowing the creation of a network that would help in the sharing of experiences and information between health professionals, in order to provide better and safer care.

This project, which was funded by the European Commission, also aids patients to understand their rights and to guide them through genetic testing and genetic counselling. *"…this project thus aims to coordinate, at a European level, a number of important aspects of genetic testing which are directly or indirectly relevant to the patient and the public."* (12)

This matter is a fairly new area which is in expansion throughout the medical field, and seeing as the evolution of the technological world has opened up opportunities in genetic testing, it is important to introduce counselling as much as possible to the population in general and it should be integrated within the health system.

Section 1.1 Elements of Genetic Counselling

As its core, genetic counselling has to focus on the main elements listed below within its practice, which should be involved in every meeting between the professional and the patient or families involved. These are as follows:

1. Psychoeducation about diagnostic and clinical aspects of genetic conditions
2. Documentation of family information and development of family tree
3. Recognition of inheritance patterns and risk estimation and assessment
4. Communication and empathy with patients and families
5. Information on available options and further measures that can be taken, in an individualised manner
6. Support in decision-making (4)

Being able to assess the risk of an individual developing a certain condition, by means of analysis of susceptibility and probabilities given by genetic testing, is one of the central aspects in genetic counselling. This assessment is to be made by the counsellor, which should then inform the patient of their results, and the adequate interpretation of them. (13)

Given that this might be an ambiguous ground, it is important that the counsellor is aware that the information given to the counselee is adequate towards their situation, and that their interpretation of it is also correct. That is why communication is such an important tool in genetic counselling. It allows the creation of a safe area to share information with the individual in question, as well as their families, in order to empower them on the best decision-making process. (4)

By means of communication, there is participation between the families, patients and the professionals in this matter, allowing a greater understanding of their genetic condition, and also, a better and easier way for communication of diagnosis between families. Communication strategies can be given by the genetic counsellor to the patient in order to make the process of communicating with relatives about a certain condition more feasible and easier. (3)

Empathic relationships are also important between the counsellor and counselee, and through means of communication, this can be achieved. Psychosocial support is a great part of the counsellors' job. By creating a good empathic relationship and having good and effective communication with each patient and their families, allows for psychological support, and also for social awareness, that can be important to take into account while counselling the patient on what options are available to them. (11)

As above stated, support is given throughout the whole process, by means of education, psychological care, and individualised guidance through treatment plans and prophylactic measures. This support should focus on the patient through the lens of the biopsychosocial model, and for that to be done it is necessary for a multidisciplinary team to be involved in this process, in order to achieve the best practice and care possible. By doing so, the patient can make the best possible decision, with all the tools and information available to him. (14)

Section 1.2 Benefits and Limitations

Genetic counselling is a new field in medical practice, and as all everything that is new is difficult to be transposed to daily practice.

At the present moment, the biggest limitation that is seen in this area is the lack of professionals that are specialised in genetic counselling, as well as the little understanding about the subject within the population in general and the health professionals. The absence of specialised education in the matter has led to a delay of its evolution and development, as professionals are unaware of the need for it. (18)

The cost associated with this practice together with the lack of understanding from health professionals leads to a lack of resources available for its development. Not having genetic

counselling, or at least primary care physicians who have been minimally trained in this area is also a great set back on introducing this preventive area of medicine. (1)

The ambiguous probabilities within genetic testing can evolve into other issues for the patient, such as psychological stress and social anxiety. A previous wholesome individual will begin to feel insecure about his health status, initiating a stressful environment in their daily life. However, sometimes the results that are elaborated formed by the professionals and given to the patients, might not even portray what is exactly going to happen in the future, as all these results are just probabilities and not an exact fact. (15)

With that being said, this issue has raised questions about psychosocial issues, bioethics, and psychosocial genetics. An example in this matter could be the carriers that are diagnosed within a family that has a genetic condition. In this case, the risk is not the same, however, it is not clear to say with certainty that this person will not develop the condition, not only due to environmental factors involved that may be risk factors associated with the condition, but also because of incomplete penetrance, or the probability of a certain condition occurring with a particular genotype, variable expressability of the mutation. (16) Therefore, in this certain scenario, the social anxiety and psychological stress caused, could be considered greater and more intense, as it remains uncertain their risk probabilities.

Being an hybrid area between psychological factors and biomedicine, has allowed individuals and families to be part of all the processes involved with genetic testing, such as diagnosis interpretation, decision-making, and others to create a deeper and more empathic relationship between the patient and the professional, which also prompts the social and psychological impact of certain diagnosis and results on the patients to be smaller and more easily overcome. (1)

Mostly all the benefits of genetic counselling fall on predictive and preventive medicine. While not disregarding the importance it has on social and psychological support, guidance and orientation, I believe the fact that it enables the prevention of certain conditions to develop is one of its main aims in conduct as well as one of the most important benefits of this area. Giving families and patients the opportunity to prophylactically treat a certain condition, or to prevent this mutation from being passed onto their offspring by prenatal diagnosis and preimplementation diagnosis, is a very beneficial part of this area.

Section 1.3 The Genetic Counsellor

The professional of genetic counselling, both medical and non-medical, should be a trained individual, at a graduate level or a masters in science (MSc) level, with capacity to analyse information, interpret results of genetic testing, have background knowledge about genetic conditions and their inheritance patterns, be able to construct a family tree for the appropriate

investigation of a familial condition, amongst other skills. However, it is important to understand that the genetic counsellor should not be the one to make a diagnosis or carry out the laboratory work that has to be involved. In fact, the genetic counsellor should carry a set of communicational and empathic skills, which will allow them to provide information to the patients in the best possible way, helping them adjust to their diagnosis, as well as informing them about their risk status, in a manner that could be understood by the patient and their families. (18)

As stated by Skirton et al, 2010, the medical and non-medical genetic counsellor must have a set of core competencies that have been developed by the Education Committee of the European Society of Human Genetics (ESHG) and other experts of the EuroGentest project, which will allow them to care for their patients in the best possible way. (19) These will make it easier to understand what their roles are and which skills they should develop and try to gain further education and training. These are as follows:

1. Establish a relationship and clarify the counsellee's concerns and expectations.
2. Make appropriate and accurate genetic risk assessment.
3. (a)Convey clinical and genetic information to counselees, appropriate to their individual needs. (b) Explain options available to the counsellee, including the risks, benefits and limitations. (c) Evaluate the understanding of the individual related to the topics being discussed. (d) Acknowledge the implications of individual and family experiences, beliefs, values and culture for the genetic counselling process.
4. Make an assessment of the counsellees' needs and resources and provide support, ensuring referral to other agencies as appropriate.
5. Use of a range of counselling skills to facilitate counsellees' adjustment and decision making.
6. Document information including case notes and correspondence in an appropriate manner.
7. Find and utilise relevant medical and genetic information for use in genetic counselling.
8. Demonstrate ability to organize and prioritize a case load.
9. Plan, organize and deliver professional and public education.
10. Establish effective working relationships to function within a multidisciplinary team and as part of the wider health and social care network.
11. Contribute to the development and organisation of genetic services.
12. Practice in accordance with an appropriate code of ethical conduct.
13. Recognise and maintain professional boundaries and limitations of own practice.
14. Demonstrate reflective skills and personal awareness for the safety of individuals and families.
15. Present opportunities for clients to participate in research projects in a manner that facilitates informed choice.
16. Demonstrate continuing professional development as an individual practitioner and for the development of the profession. (19)

The genetic counsellor is usually seen as a non-medical professional, in other words, not a doctor *per se*. However, these professionals can also be genetic specialists that have had such experience in this area, as well as the possibility of having some kind of specialised formation. Nevertheless, sometimes genetic specialists may lack additional education in order to gain certain skills and aptitudes that are needed in this context. Thus, making it important for the

introduction of a genetic counsellor that has already had that education, and has acquired these sets of skills. (20)

It is important to understand that the genetic counsellor should be integrated into a multidisciplinary team that includes doctors, nurses, psychologists, social workers, and any other professional that would seem fit in each patients' case. These teams can deliver genetic counselling services in a more effective way, following the biomedical model of practice, and approach the patient and families in a holistic manner. (11)

Chapter 2: Oncogenetics and Genetic Counselling

Hereditary cancer syndromes have a great impact on patients and families that have to endure this diagnosis. A prompt diagnosis provides the opportunity to undergo preventive measures as well as cancer screening and treatment that will eventually minimise the impact of this condition. (21) Cancer genetics allows for surveillance and early diagnosis of these conditions in certain individuals.

There are four main groups that are considered in oncogenetic counselling, and which the counsellor should be familiar with and able to inform their patients and families on what the best arrangements, for each individual situation are. These are as follows:

1. familial tumour syndromes following mendelian (usually but not always autosomal dominant) inheritance;
2. common cancers of later life and their genetic subsets;
3. genetic disorders giving a general predisposition to malignancy (commonly autosomal recessive);
4. embryonal and childhood cancer.(4)

As cases of mendelian inheritance, the involvement of relatives is extremely important. By mendelian inheritance, what I mean is that cancer syndromes are examples of disorders inherited according to Gregor Mendel's laws, with a pedigree compatible with a specific type of inheritance, usually autosomal dominant, in which the risk to offspring of affected members will be one-half. (4)

An important minority of these cases follow this kind of inheritance pattern, however, even though it is a minority, it implies several families that should be cared for. (4) This caused health professionals, in general, to change their attitude towards genetic services and to be more aware of which families might need this kind of management.

As its main purpose, oncogenetic counselling focuses on the following points:

- Allow individuals to understand their condition, their risks of developing certain types of cancer, and what management options they have available to them.
- Support diagnosis and preventive measures of genetic diseases.
- Guide patients through the decision-making process, as well as aid with communication strategies to address this subject with their families. (site APPAGEN)

In oncogenetic counselling it is important to identify the index case and which families follow mendelian inheritance patterns, and in this particular case, these will benefit from genetic testing in order to identify a germline mutation. Genetic counselling should be carried out in all family members, including asymptomatic family relatives, when a familial mutations is identified. PST could be option to asymptomatic family relatives when the familial pathogenic variant is identified, which can only be prescribed by medical geneticists. In these cases it is also important to inform about the reproductive options for young couples, namely the prenatal diagnosis and the preimplantation genetic diagnosis. (11)

These objectives do not differ greatly from the core competencies in general, however, oncogenetic counselling is a branch of genetic counselling and has different implications to it.

The genetic counsellor should be adequately trained to deal with these patients and their families and by so, be able to collect the necessary family information and mutation tracking information to create a pedigree-based study. The counsellors should be aware that this kind of disorder can be associated with a social stigma that can make this process harder for the patient and families involved. (10)

Given the wide variety of technology offered to the population, and the fact that it is possible to have direct-to-consumer genetic testing, without any counselling, emphasises how important the implementation of genetic counselling really is. The perceived risk of hereditary cancer that can be deduced by personal genomic testing, can cause changes to risk behaviours that might even be unnecessary for that individual. (22)

Oncogenetic counselling not only includes the patients and families that are diagnosed with a mendelian subset, but also all the patients that suffer from oncological conditions, and that are candidates for genetic testing or genetic counselling. Even though a patient does not have a family history of cancer, maybe a genetic testing could be considered due to the risk that falls on the offspring.(4)

Also, it is important to understand that oncogenetic counselling should be included in the skill set of health professionals, especially at a primary care level. These doctors and other professionals should know which patients to refer to genetic counselling, and also they should be able to apply some of the counselling techniques and skills in order to understand if that patient is a candidate for a referral or not. These skills are very important and should be implemented at a primary care level, in order to more easily refer the adequate patients, and by this initiate and apply preventive medicine management. (23)

According to Jhaveri et al, the following table summarises the criteria that should be met when deciding which patients should be referred to oncogenetic counselling.

Table 1: General criteria for referral of patients to cancer genetics services (21)

A personal or family history of early-onset cancer (eg., younger than 45 years for breast cancer, younger than 50 years for colon or uterine cancer)
Multiple family members on the same side of the family with the same or related cancers
A family member with a diagnosis of more than one type of cancer
A personal or family history of breast, ovarian, or pancreatic cancer who are of Jewish ancestry
A personal of family history of a rare type of cancer/tumour (eg., breast cancer in a male, medullary thyroid cancer, a sebaceous carcinoma or adenoma

Section 2.1. Hereditary cancer syndromes

There are various hereditary cancer syndromes, and many still have molecular research being carried out. Even though there are many genes identified associated to these conditions, therefore there is room for oncogenetic counselling practice, as there are patients that undergo genetic testing.

According to their prevalence, hereditary breast and ovarian cancer (HBOC), Lynch Syndrome (LS) and familial adenomatous polyposis (FAP) are syndromes that have been studied in greater depth in relation to the subject of this dissertation. Seeing as they have mendelian inheritance, they are common examples of hereditary cancer syndromes observed at oncogenetic counselling.

HBOC, which has an autosomal dominant inheritance pattern, has a prevalence of 5-10% (24), and the genes that are involved in this syndrome are *BRCA1* and *BRCA2*, being these the ones considered the high penetrance genes (25), with higher risks of susceptibility of developing breast or ovarian cancer (26). American and European recommendations for oncogenetic counselling referral are based on the presence of *BRAC* genes in family history or patient's medical history, such as the history of early onset breast cancer, or two or more primary breast cancers in the same person.

FAP is caused by a mutation in the *APC* gene, which leads to almost 100% lifetime risk for individuals that carry this mutation. (7) Again, and as is the case of breast cancer, referral is also based on the presence of the mutation as well as family history.

In regards to (LS) which is associated with four different mismatch repair (MMR) genes, *MLH1, MSH2, MSH6* and *PMS2*, and a non-MMR gene, *EPCAM*. This syndrome is responsible for 2-5% of all colorectal cancer cases, where *MLH1* and *MSH2* account for 90% of pathogenic variants identified in families with LS.(27) This syndrome has the Amsterdam and Bethesda criteria, which dictate correct risk assessment, referrals and management strategies. However, these have been undergoing revision. (7)

The tables below show the values of susceptibility that the population with HBOC and LS that has these mutations is under in comparison to the general public. It is important to point out that both these syndromes can give rise to other types of cancer, for instance, HBOC can be the cause of prostate cancer, while LS can cause gastric or ovarian cancer.

Table 2. Cancer risk comparison of individuals with LS aged ≤70 and the general population (27)

Cancer Type	**General Population Risk**	**Lynch Syndrome (*MLH1* and *MSH2* heterozygotes)**	
		Risk	**Mean Age of Onset**
Colon	4.8%	52%-82%	44-61 years
Endometrium	2.7%	25%-60%	48-62 years
Stomach	<1%	6%-13%	56 years
Ovary	1.4%	4%-12%	42.5 years
Hepatobiliary tract	<1%	1.4%-4%	Not reported
Urinary tract	<1%	1%-4%	~55 years
Small bowel	<1%	3%-6%	49 years
Brain/central nervous system	<1%	1%-3%	~50 years
Sebaceous neoplasms	<1%	1%-9%	Not reported

Table 3: Comparison of risk of malignancy in the general population and carriers of germline *BRCA*1 and *BRCA2* pathogenic variant (28)

Cancer Type	General Population Risk	Risk for Malignancy	
		BRCA1	***BRCA2***
Breast	12%	46%-87%%	38%-84%
Second primary breast	2% within 5 years	21.1% within 10 yrs 83% by age 70	10.8% within 10 yrs 62% by age 70
Ovarian	1%-2%	39%-63%	16.5%-27%
Male breast	0.1%	1.2%	Up to 8.9%%
Prostate	6% through age 69	8.6% by age 65	15% by age 65 20% lifetime
Pancreatic	0.50%	1%-3%	2%-7%
Melanoma (cutaneous & ocular)	1.6%		Elevated Risk

These are just examples of cancer syndromes, seeing as these are the ones where there is more literature about, and a fair share of the investigation. Other common cancers that may also have genetic implications are oesophageal, gastric, ovarian and endometrial cancer. (4)

Section 2.2 Risk assessment and oncogenetic susceptibility

Risk is always an ambiguous ground, as it is not an exact and precise value of what will happen in the future. "*Information on genetic risk is rarely an absolute yes or no (…) one works almost entirely in terms of probabilities or odds.*" (4) This makes risk assessment a challenge that needs to be clearly evaluated by a trained professional so that the correct information and orientation of patients and families is given.

It is possible to say that mendelian inheritance is associated with high risks, however, the majority of cancer cases showcase a weak inherited influence. This shows that in the area of cancer genetics, there is the need to understand which type of inheritance, if it is autosomal

dominant, autosomal recessive or X-linked, and the nature of the disorder. This influences risk assessment and susceptibility of individuals. (4)

Firstly, in order to evaluate risk, it is important to have a good clinical assessment of the patient, as well as the adequate information that can influence risks, such as family history, the age of onset, and type of cancer. Also, making a pedigree analysis is crucial to understand inheritance patterns. After gathering information about the patient and respective family members, genetic testing comes in for the identification of genes and mutations that can be involved in these conditions. For example, if a patient comes in with the history of breast cancer at age 25 and positive family history, the genetic testing will be aimed at the identification of genes involved in breast cancer. Therefore, counselling prior to genetic testing can also aid in orientating the course of genetic testing. (25)

Factors involved in the stratification of risk, such as the age of onset, family history, tumour details, amongst others can be used to develop guidelines for the assessment of patients with certain conditions. For instance, in colorectal cancer, the Amsterdam and Bethesda criteria can be used to assess risk and to easily categorise patients into different risk groups, and with that follow specific guidelines for each group. (4)

Therefore, risk assessment requires information that has to be gathered by different specialities in medicine, such as surgeons, primary care doctors, radiologists, oncologists, as well as the genetic counsellor and a medical geneticist. Given this, these professionals should be aware that teamwork is essential with these patients and families, in order to provide the adequate information with the correct indication for future management according to their risk stratification. (4)

Problems for risk assessment are the presence of incomplete penetrance and variables of uncertain significance. (17) This creates a group of individuals which can be at risk, however, the uncertainty rises due to the phenotypic spectrum that can arise from this kind of genes. (29)

Given the increase of investigation to these syndromes, Slavin et al proposed the following Table 4 as a way to categorise into risk groups the genes involved in each condition, in order to manage these patients in the best possible way, seeing as risk stratification has implication on the future conduct necessary for each patient. (26)

Table 4: Breast, colorectal and ovarian cancer risk estimates (26)

Cancer Site	**High Risk (odds ≥ 5,0)**	**Moderate risk (≥2,0 odds < 5,0)**	**Low risk (≤2,0 odds ≥1,o or growing evidence of association**
Breast (female)	BRCA1, BRCA2, CDH1, PTEN, STK11, TP53	ATM, BRP1, CHEK2, PALB2	BAP1, BARD1, RAD50, RAD51C, RAD51D, MRE11A, MUTYH, NBN, XRCC2
Colorectal	APC, BMPR1A, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, SMAD4, STK11	CHEK2, PTEN, TP53	CDH1, EXO1, GALNT12, MUTYH, POLD1, POLE
Ovary	BRCA1, BRCA2, MLH1, MSH2, STK11	MSH6, PALB2, RAD51C, RAD51D	BARD1, BRP1, CHEK2, MRE11A, MUTYH, NBN, RAD50, TP53

The same author proposed figure 1 as a way to understand the clinical utility that can be provided to each group. It shows that the higher the risk stratification, the more clinical utility these patients have. By clinical utility, the author meant, how much can be done for these patients at a clinical level, such as preventive treatment or management. This figure also has information about each risk group, and together with the Table 4 can better identify susceptible individuals. (26)

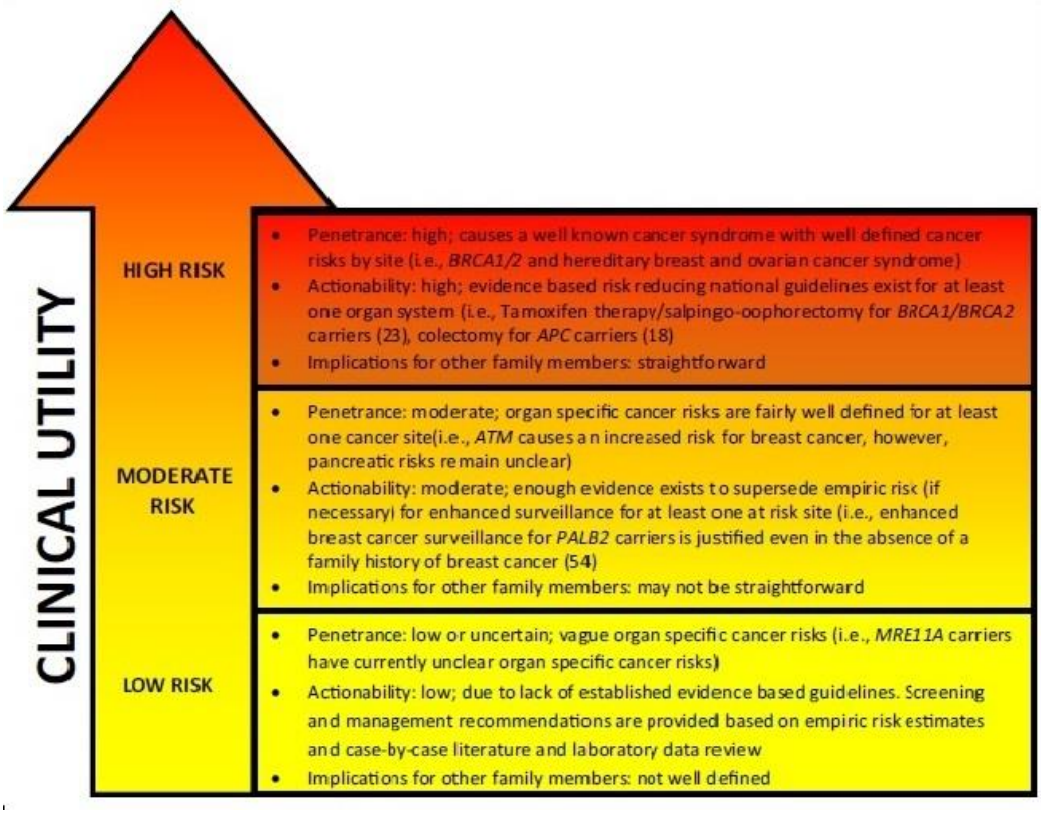

Figure 1. General characteristics of genetic cancer risk groups and their clinical utility.

When talking about risk, we need to take into account how this is interpreted by the patient and families. This is of great importance because this interpretation can affect their psychological status and ultimately their involvement in this process. It has been stated that 25% of patients are aware of their real risk status, while 50% overestimates their risk status. (30) Due to this overestimation, it is important that the genetic counsellor is able to correctly communicate what each risk status indicates and implies. (18)

Section 2.3 Ethical Considerations

Since the beginning of genetic testing, and the discovery of the technology that can be used towards the development of genetics, many ethical issues have been raised. These should be considered in respect to the bioethical principles of justice, nonmaleficence, beneficence, and autonomy. The duty to inform a patient that is put upon the health professionals according to their code of conduct elaborated by ethics committees should always be considered by the counsellors, in order to provide the best care for their patients.

In order to more easily describe the ethical considerations necessary in each principle, I will focus on each principle individually.

In view of the principle of autonomy, the patient is entitled to decide on the best course of action towards its own genetic material. The counsellor should only advise the patient and their

families on what the best strategy would be for their specific case, however, the ultimate decision relies on the patient in question. Giving the patient the proper information prior to any genetic testing is key for the patient to be able to make a fully informed decision, considering all their beliefs and convictions. (31)

The principle of beneficence focuses on the benefits of genetic testing and counselling. By providing a psychosocial well-being of the patient by means of counselling, the patient can then make better decisions for the future, and arrangements for what can be seen as necessary.

Privacy of results and diagnosis, as well as all the psychological implications for each patient and their families, should be kept private from third-parties, to avoid discrimination and stigmatisation. By doing this, psychosocial risks are diminished and this is very important in terms of nonmaleficence. (32) In this principle, it is also important to understand that the genetic test are not compulsory, and the patient and families can decide whether or not to undertake them. (31)

Finally, the equal access of the population to these services is crucial. This allows all the individuals at risk to undergo the correct testing and counselling. It also lets the health professionals to correctly identify the individuals that need further testing and the ones that do not. This has been seen as a challenge due to the lack of professionals in this area, especially fully trained genetic counsellors. (31)

Chapter 3: The Portuguese Scenario

In 1998 the medical speciality of Medical Genetics was recognised by the Portuguese association of medical doctors (Ordem dos Médicos – OM) and becomes an acknowledged and integrated speciality in the National Health System (NHS), initiating the first internship in 2002. (31)

However, less than 50% of clinical geneticists in Portugal are practising. (32) Most of them work at a laboratory level, and others mainly work by themselves in a private office, with no use of a multidisciplinary team. In Portugal, the clinical practice of genetic counselling is mainly given by oncologists or medical geneticists. (33)

By 2015 there was 53 medical genetics registered at the OM, and there are two non-medical genetic counsellors integrated into the NHS. Non-medical genetic counsellors are still unrecognized professionals, which is why it is hard for the incorporation of genetic counsellors in the NHS. (33)

According to Paneque et al, in 2015 Portugal had five medical genetics services as illustrated in the figure below. As it can be seen, these services are mainly in the two major Portuguese cities, Lisbon and Porto, and the one medical service located more to the east of Portugal is now seriously understaffed. (33) One of the Portuguese objectives is to have ten genetic services integrated into the NHS by 2024. (31)

There are also regional oncological hospitals, *Instituto Português de Oncologia* (IPO), that are a part of the NHS where they have medical genetics services, more precisely, familial risk consultations and oncogenetic. There are only two of these hospitals in Portugal, one in Porto and one in Lisbon. (31)

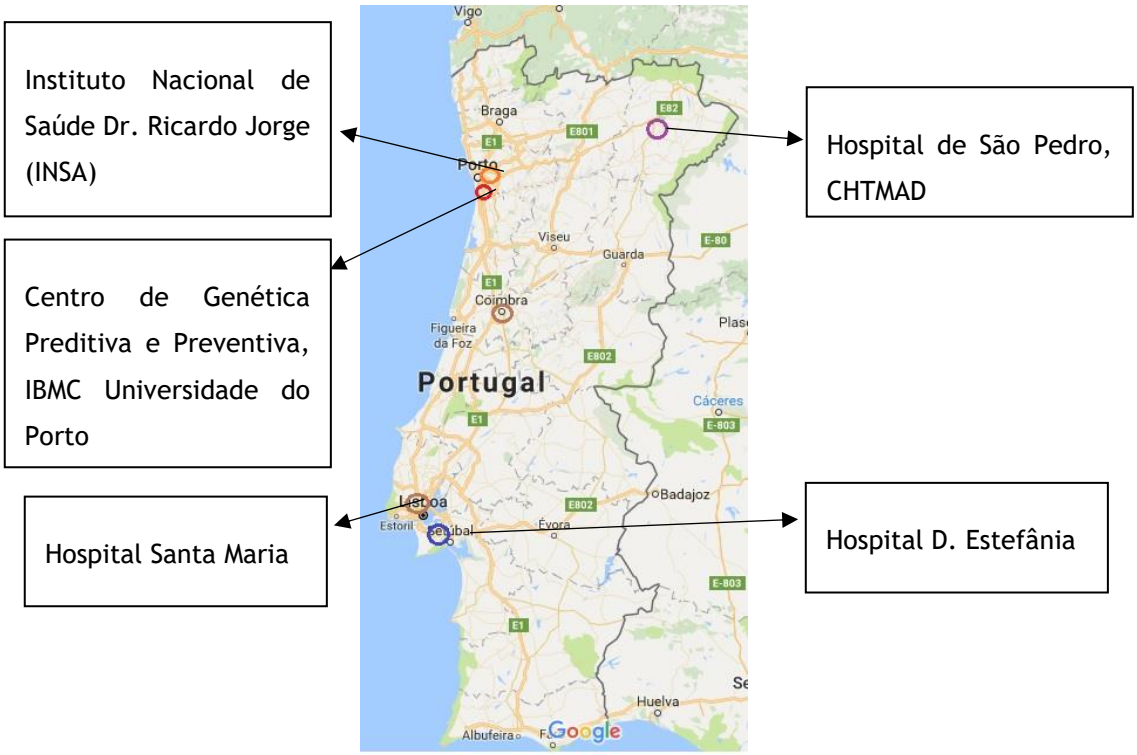

Figure 2. Map illustrating the location of Portuguese genetics services.

Table 5: Portuguese Institutions with genetic services available within the national health system (31)

Region/Institution	Clinical	Laboratory
North		
Instituto Nacional de Saúde Dr. Ricardo Jorge (INSA)	Yes	Cytogenetics Molecular Genetics Biochemistry
Centro Hospitalar de Vila Real/Peso da Régua	Yes	Cytogenetics
Centre		
Centro Hospitalar de Coimbra – Hospital Pediátrico	Yes	Cytogenetics
South		
Hospital de Santa Maria	Yes	Cytogenetics
Hospital Dona Estefânia	Yes	Cytogenetics
Hospital de Egas Moniz	Yes	Cytogenetics

Cancer genetics services are integrated into the NHS as part of general and oncological hospitals. These services have the help from national laboratories that are also partly integrated into the NHS. There are also many private laboratories and practices that work in parallel to the NHS. This also allows for more scientific investigation and the possibility to enhance the genetics services. (33)

An example of such laboratories is INSA, which is a public organisation that is under the tutelage of the MS, and it is considered the main laboratory system in the NHS. This institute is part of

many national and international organisations, such as the Centre for Disease Control at an international level, and *Instituto de Biologia Molecular e Celular* (IBMC) at a national level.

This institute has a human genetics department where it focuses on genetic testing, and investigation of scientific as well as technological improvements for this kind of testing. This institute could provide genetic counselling services, as it provides genetic testing for numerous of areas, such as pre-natal diagnosis, reproductive purpose, amongst others. (34)

The IBMC is an important link between the university researchers and health professionals that work within the hospital services. This is mainly a multidisciplinary research institute as it involves many areas of medicine. However, in the area of genetics, IBMC has a department which is the *Centro de Genética Preditiva e Preventiva* (CGPP), where it carries out genetic testing, and genetic counselling for patients and their families who might be indicated to do so. (35)

At a private level, there is the *Centro de Genética Clínica* (CGC) which is the main genetics laboratory in Portugal and the leader of genetic testing. This centre is also one of the main laboratories at a European level, due to their development of exclusive technology for genetic testing. (36)

Portugal has a national association of genetic counsellors (APPAcGen), which is a non-profit organisation with scientific, cultural and social purposes for the improvement of the practice of genetic counselling practice taking under consideration scientific, technical, organisational, ethical and human issues involved. The current president is Dra. Milena Paneque, a pioneer in genetic counselling at a national level. This association has helped the implementation of genetic counselling in the Portuguese health services, as well as raise awareness for the need to integrate more genetic counselling professionals within the NHS in order to enrich the services provided to patients and families undergoing genetic testing. (37)

Section 3.1 Portuguese legislation for genetic testing and genetic counselling

National legislation concerning genetic testing was elaborated in 2005, Law nº 12/2005, which has been reviewed in 2016, states that presymptomatic, carrier and susceptibility testing, as well as prenatal and preimplantation diagnosis, should only be carried out after appropriate genetic counselling and written informed consent. It also stresses that whenever an individual or family are diagnosed with and hereditary condition, these should be referred to genetic counselling services. (38)

The Ministry of Health (MS) stated that this counselling should be given by a clinical geneticist or any other physician. (39) However, given the fact that genetic counselling is still scarce in

Portugal, the amount of clinical geneticist is insufficient for this, and the other physicians are not adequately trained. (33)

The *Conselho Nacional de Ética para as Ciências da Vida* (CNECV) is an independent organisation which aims to analyse ethical issues within the biomedical field. This organisation highlights the need for genetic counselling for patients that are undergoing genetic testing or are seemed fit for genetic testing. By means of conferences, seminars and reunions, it has been able to raise awareness to the ethical issues involved within the genetic field, and how these should be overcome and what is necessary to be done by health professionals in order to pass on information to patients and their families in an ethical and responsible way. (40)

Section 3.2 Referral network in Medical Genetics

A study carried out by Leandro et al stated that 68% of the physicians involved in their study had never referred a patient for genetic counselling and that the main professionals that do refer their patients are primary care doctors (40%) and internists (33%). Even though this study is based on hemochromatosis referral in Portugal, it still indicates how this referral network is poorly used by the Portuguese health professionals. This is due to the lack of understanding of genetics and also due to the lack of knowledge of which services are available for patients. (41) These results show the need for a referral network to be implemented and for clinical physicians to be aware of it.

This network has been developed by the General Health Directory (DGS), in 2014, and it aims to articulate all medical specialities with medical genetics, create opportunities for better understanding of the resources available at the NHS level, and also to investigate and study what can be done to improve the access to these services by the general population. (31) By means of this referral network, there were objectives that were proposed by the DGS in order to improve the medical genetics services, such as the implementation of more medical genetic services in the NHS for better equity of access by the population.

The referral network report can also be used as a guideline for other clinical physicians, such as primary care doctors, on what to do when referral of these patients to a medical geneticist is necessary. This allows the Portuguese health professionals to be aware that there are these services available in the NHS and gives them an orientation on how and when to use this network. (31)

Section 3.3 Graduate education in genetic counselling

In 2009 at the *Instituto de Ciências Biomédicas Abel Salazar* (ICBAS), which is part of the Porto University (UP), a professional master's program in genetic counselling was initiated. In 2011 the first class graduated with six students, three nurses and three psychologists. Two of them

have integrated into the NHS, one is working at an oncological hospital and the other three have returned to their previous jobs. (33)

This MSc course consists of a two-year full-time programme, with a curriculum based on the core competencies of genetic counselling, that were developed by the ESHG, as stated above. It is known that this MSc program was not available in 2012/2013 and 2014/2015, however, I was unable to understand if it is still available at the present time. (33)

This master programme was accredited by the ESHG, and its main objectives are to adequately train students in genetic counselling, equipping them with the necessary skills for clinical practice and scientific investigation, as well as promote awareness of the need for multidisciplinary teams in this matter and the psychosocial impact this can have on patients and their families. (32)

This master's programme is the sixth of its kind in Europe, which means that more and more training of genetic counsellors is becoming generalised, allowing for the development of a good inter-professional network at an international level. This also permits professionals to share their experiences and to carry out investigations based on a wider population base. (33)

Chapter 4: Portugal and Europe

As a member state of the European Union, Portugal has obviously been compared with other European countries in relation to many issues. It does not differ in the matter of genetic counselling and oncogenetic counselling.

Many European countries have started developing their genetic counselling training and legislation and initiated to integrate these services within each countries NHS. (20)

The ESHG aims are to promote research in basic and applied human and medical genetics, to ensure high standards in clinical practice and to facilitate contacts between all professionals who work in this area, especially in Europe. The society encourages and seeks to integrate research and its clinical benefits onto the professionals and public education in all areas of human genetics. (42)

This society includes committees and subcommittee that focus mainly on an area of human genetics, as is the European Board of Medical Genetics (EBMG), which aims to provide the best clinical practice to patients that are looking for medical genetics services. It focuses on three professional branches, genetic nurses and genetic counsellors, clinical laboratory geneticists and medical geneticists. The EBMG has supported genetic counselling in Europe by supporting educational programmes, registration of genetic nurses and counsellors, as well as stimulating the development of standards of competence. (43)

The branch board of Genetic Nurses and Genetic counsellors (GNGC) in EBMG, allows for the registration of genetic counsellors, following a rigorous registration process, where professionals need to be eligible to register by having the appropriate training in genetic counselling or as a genetic nurse. In this manner, it is possible to gather all these professionals at conferences for experience sharing, training expertise, and knowledge about the matter, and also to create a safer practice of the patients seeking these services. (42)

The registration of the GNGC shows that there is only one Portuguese registered, which is Dra. Milena Paneque in 2015. The European country with the most number of registers is France, followed by Spain, the UK and Norway. It is important to state that the registration includes both counsellors and nurses, therefore, it is not certain that the numbers of registered professionals are genetic counsellors. (42)

However, there is a clear discrepancy on how to manage these patients throughout the European countries, and it is still unclear on which is the best way to practice genetic counselling. There is still a challenge on finding the most effective counselling practice, and it is important to do so seeing as the effectiveness of counselling comes from the process itself.

This calls for the development of guidelines and protocols that can be used as a framework, to help genetic counsellors around Europe manage their patients. There is also need for the development of quality assessment tools for genetic counselling, that has been shown by Paneque et al on a Delphi study carried out in European countries. (44)

In terms of genetic testing, and as already stated above, the EuroGenTest was established to help European countries follow the same referral criteria, and aid each other with epidemiological numbers in order to understand what areas are lacking investigation or need to be further investigated. This platform allows for consensual managing skills and competencies, allowing all patients to have an equal and safe care provider. (12)

As above stated, Portugal already has an MSc program in genetic counselling, following other European countries, such as the UK, Spain, Russia, Germany, amongst others. However, these master programmes do not follow the same curricula, causing great variation in the training they provide, showing that there is still need for consensual information throughout Europe, and there is a need for societies such as ESHG and EBMG. This program is accredited by the ESHG (5), which allows for future trained professionals to be accredited at a European level.

In regards to legislation, most European countries who practice genetic counselling have some form of legislation, at least about genetic testing, where genetic counselling is briefly talked about, as is the case of Portugal. France and Norway have specific legislation for genetic counselling, while in Iceland, Turkey, Sweden and the UK, genetic counselling would be subject to statutory regulation. In France, The Netherlands, Norway and Sweden, it is illegal for non-medical genetic counsellors to work without a medical doctor. (20)

However, not all countries have this kind of legislation, for instance, in the UK there is no legislation related to genetic testing in general, whereas in France the legislation is strict about genetic testing and it takes into consideration genetic counselling. Again, showing a discrepancy in the legislation of genetic testing, genetic counselling and other genetic related matters throughout Europe. (45)

As a small comparison, and by the numbers given by the GNGC, it seems Portugal still has to evolve in this area and integrate non-medical genetic counsellors into its health services. Nevertheless, it is walking in the right direction and soon will reach the standards of other European countries.

Conclusion

Oncogenetic counselling is an important aspect of the oncological patient and their families. Counselling allows for a better adaptation to normal life after diagnosis or even during PST, enabling patients to easily overcome all the psychological stress associated with this in a more calm and orientated manner. Also, the fact that the patients and their families are aware of their condition and what it implies on their personal and professional life, can also give them a sense of empowerment that could help them overcome their issues, and allow them to have a better psychological state of mind to carry out their decisions. (8)

Health professionals that follow these patients should be an active part of a team, with a proactive attitude and ready for inter-professional education and training. The multidisciplinary teams are the pillars of genetic counselling, and by working together they will provide the best possible care for the patient and families. These teams have shown that genetic counselling becomes more effective, with better responses from the patients that have to undergo this. (33) Integrating primary care physicians into these multidisciplinary teams would be advantageous, seeing as these are the medical professionals that have the greater contact with the patient and their families, and also are the professionals that are more aware of the social and psychological status of these patients. Training primary care doctors to be aware of inheritance patterns, and to know when an individual can be at risk, will allow for earlier referral, hence earlier management and investigation.

Having a non-medical genetic counsellor would be able to help out the clinical geneticists as well as other physicians in the best way for communication, counselling and support of these patients and their families. (18) However, it is necessary to improve training of physicians in this area, as well as adequately inform them of referral networks, in order to effectively care for these patients. (46)

At a national level, genetic counselling is carried out by medical geneticists, and some services already include this practice. However, Portugal still lacks protocols and guidelines that are adequate to the cultural needs of the population, and this makes it harder for the professionals involved to carry out their duties in a consensual manner. This can raise doubt within the patient and their families, as they can be given different information from different physicians or non-medical genetic counsellors.

Also, the lack of resources, databases and services available for the development of this area, together with the little awareness and non-specialized training within the medical population creates a barrier for adequate referral of these patients, and this needs to be improved. Non-medical genetic counsellors are still not integrated in the NHS, which should be considered as a future method to try to develop this practice in Portugal.

There is a need for inter-professional and inter-institutional networks involved in cancer genetic care at a national and international level (9). These networks, that are already beginning to appear at a European level, are crucial for the correct conduct of genetic counselling, as well as a good manner to elaborate guidelines and protocols for each individual condition that is addressed. By experience, these professionals can help each other provide the best care for the patient, always considering the biopsychosocial model.

As it is seen in Portugal, many European countries have very few non-medical genetic counsellors, and have this practice be carried out by oncologists and medical geneticists, while others, such as the UK, have integrated non-medical genetic counsellors within their NHS. Having an international network will help for this integration to happen due to the sharing of experiences by these countries.

This dissertation, as a literature review has the limitation of not having direct sources of information. However, by the literature reviewed it is feasible to conclude that oncogenetic counselling in Portugal is still in its prime years, even though there have been considerable advances in terms of medical genetics and recognition of the genetic counselling profession.

Future Perspectives

As a still expanding area of medicine, genetic counselling still has a long way to grow, especially in Portugal. The fact that there is already a master's program and legislation concerning this matter, allows for Portugal to be in the right direction, and following the right path for total implementation of these services in the NHS and raise awareness to their importance and necessity.

Even so, it is important to try to come up with strategies to overcome the difficulties seen in genetic counselling. After reviewing the literature about this matter, the strategies I propose are as follows:

- Create more graduate programs and encourage training of primary care physicians
- Create a database that can help with referral of patients
- Integrate genetic counselling in the NHS and recognise genetic counselling as a health profession
- Create databases and reports about genetic conditions in order to have more information about these subjects available to physicians
- Raise awareness within the population for the importance of genetic counselling, as well as the services available to them by the NHS, so that individuals and families can also take part in this process and reach out for help if necessary.

The population, including doctors, might not be aware that the NHS provides these services. I believe that by raising consciousness about this subject could aid the development and recognition of the genetic counselling.

Europe has already come together in this area and is beginning to develop consensual guidelines for genetic counselling, encouraging other countries that still have not developed this area to do so, and also, this brings about a great inter-professional network at an international level that will allow genetic counsellors to continue their training and education by learning from other colleagues around the world.

In order to continue this development, it is important to bear in mind these key questions and to try to overcome them in order to succeed.

1. How to raise awareness within the population about the role of genetic counselling and the genetic counsellor?
2. Are there psychosocial models of genetic counselling that are based on the cultural needs of the Portuguese population?
3. How to implement psychosocial and educational evaluation programs?
4. How to facilitate the process of genetic counselling?
5. What investigations based on evidence have been published that can be used to enrich the quality of genetic health care?

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