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RESEARCH

Service provision of genetics health care in Portugal

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Abstract
In recent decades, genetics has undergone important technological advances. The rapid shift to genomics has made a strong impact on health systems around the world. In Portugal, this huge increase in consultations and typologies of genetic tests has joined the serious limitations of the few existing genetics services. The following study aims to characterize the current state of the network of genetics services in Portugal regarding its functioning, main challenges, and opportunities. Five semi-structured interviews were conducted, corresponding to 83.33% of the directors of the public genetics services of the National Health Service. Four thematic categories emerged from the analysis: (1) specialty and technical developments, (2) structural difficulties, (3) potentialities, and (4) future directions. The developments are due to the emergence of more comprehensive genetic applications, specific protocols and patient referral standards, and accreditation of services. The main difficulties encountered in the functioning of the services were difficulty in obtaining funding, lack of human resources, service overload, and lack of exclusive time for training and research. The potentialities mentioned were the establishment of multidisciplinary teams and the best articulation with specialists from other areas. Among the various future directions pointed out, better management of patients' waiting lists, the importance of research, the simplification of test request procedures, and the creation of specialized units inside the genetic services, were reported. The results showed several gaps in the practice of medical genetics that should be addressed with the development of public policies for the recognition and restructuring of medical genetics in health care.

Keywords Genetics services · Genomics · Health policy · Health services accessibility · Human genetics · Public health

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Introduction

Genetics, as a scientific discipline, originates between the 6th and 8th decades of the nineteenth century (McKusick and Harper 2013). Medical genetics services are part of health institutions and are dedicated to the diagnosis, treatment, follow-up, and counseling of people and their families, as well as carriers or at risk of being carriers of genetic diseases (Epstein 2006). Historically, genetics services were isolated in tertiary care environments, generally in large academic centers (Battista et al. 2012).

In the context of medical genetics in Portugal, the study of genetic diseases began in 1939 with the diagnosis and evaluation of family diseases with onset in adulthood (Saraiva et al. 2001). Subsequently, karyotype analysis emerged in 1959, and the teaching of genetics emerged from 1970 onwards, with the creation of the first genetics course at the University of Porto (Universidade do Porto), initially as part of medical pathology and as an autonomous discipline since 1981 (Harris et al. 2000).

In 1979, there was the creation of the Medical Genetics Competence Commission (Comissão de Competência de Genética Médica), and the following year, neonatal screening tests became available in Portugal (Santos et al. 1997). The hospital career was established in 1982, and in 1983, the Medical Association (Ordem dos Médicos) recognized medical genetics as a specialized area (Saraiva et al. 2001).

The first cycle of special studies in medical genetics was created in 1986 at the Center for Medical Genetics Doctor Jacinto Magalhães (Centro de Genética Médica Doutor Jacinto Magalhães). In 1999, medical genetics was officially accepted as a specialty, and the College of Specialty of Medical Genetics (Colégio da Especialidade de Genética Médica) was created. The specialty was recognized by the European Union on 4 March 2011, making it a recent medical specialty in Portugal (Saraiva et al. 2001).

Despite being a new specialty, with the rapid expansion of latest generation sequencing, accessibility of testing, and the public interest, genetics is increasingly relevant in many areas of medicine and health (Harding et al. 2018). Precision medicine's vision carries the promise of transforming health care, addressing serious gaps in the provision of genetics services (Flannery 2018).

One of the clear advances is the significant improvement of genetics diagnosis. In recent years, constant advances in technology have given rise to next-generation sequencing, enabling genetic analysis at different levels, including gene panels, exomes, or genomes (Buermans and Dunnen 2014).

In addition, the evidence base demonstrating that genetics counseling produces positive results for people with common complex diseases is expanding (Madlensky et al. 2017). In recent years, rare diseases have been taken into account at all levels of government, health, and patients, which has strengthened the role of medical geneticists in multidisciplinary teams (Alonso et al. 2011; Baldovino et al. 2016). There is also a relevant role in public health, since diseases represent a significant problem because of the burden that disease is for society as a whole—involving family members, caregivers, health professionals, and the community.

However, the current knowledge about health services in genetics in Portugal is quite insufficient, and their status is not as encouraging as in some reference countries. Thus, over the last few years, multidisciplinary work has been developed for the evolution of genetics, genetic counseling, and the identification of structural and human insufficiencies (Paneque et al. 2014, 2018, 2021). To better understand the configuration of National Health System (SNS) and its influence in genetic services structure, it is important to highlight that SNS is composed of five Regional Health Administration (ARS) units—North, Center, Lisbon and Vale do Tejo, Alentejo, and Algarve—that establish the contact between the Ministry of Health and local structures. Considering funding, public genetics services are part of the SNS; thus, all professional salaries, consultations, and genetic tests are paid with public funding (Law No. 95/2019, September 4th).

In turn, genetic counselors are not yet recognized in Portugal as professionals by the competent national authorities. Once the large pharmaceutical companies do not present a specific interest in genetic services, the expenses with training or scientific conferences must be covered by the genetic professionals themselves. Last, concerning genetic tests if they are carried out in public services, all costs are covered by SNS; however, the waiting time for consultations and test results are often longer than in private services. Sometimes, SNS assures the costs of genetic tests in private services through an agreement with ARS. The management of financial and human resources, installations, and equipment of the SNS, as well as to the definition and implementation policies, standardization, regulation, and planning in health, in articulation with the ARS is made by the Central Administration of the Health System, I.P (ACSS, I.P.).

Identifying the necessary structures for the provision of genetics services and the indicators of quality and effectiveness of the care provided in them requires the definition of the services' objectives (Kaye et al. 2020). The aim of this study is to map the Portuguese medical genetics services, specifically (1) its functioning, (2) the main challenges, (3) current opportunities, and (4) future opportunities, in order to contribute to the reorganization of this essential area in health care.

Material and methods

For this exploratory descriptive study on the characterization of the network of genetics services in Portugal, we have opted for a qualitative methodology, through semi-structured recorded and transcribed interviews. The qualitative approach provides a deeper and more comprehensive perspective on the theme, particularly relevant when it is intended to generate and deepen the contents related to a particular line of research (Mcleod 2001), in a contextualized way. The five directors of the country's public genetics services and three directors of genetics units of the National Health Service (Serviço Nacional de Saúde) were invited to participate—one of them in the process of obtaining the status of service. Thematic analysis was used with the aim of broadening knowledge on the subject, through the identification and analysis of meaning patterns from qualitative data (Gubrium and Holstein 2001; Mcleod 2001; Braun and Clarke 2006). The study had the positive feedback and approval of the Committee for Ethics and Responsible Conduct (Comissão de Ética e Conduta Responsável) in the research of the Institute for Research and Innovation in Health (Instituto de Investigação e Inovação em Saúde).

Participants

Four genetics service directors participated in this study and additionally one director of genetic consultations in the process of obtaining service status, whose testimonies represent 83.33% of the global reality of the country's public genetics services. The interviewees were the directors of the genetics service of the University Hospital Center of Coimbra (Centro Hospitalar Universitário de Coimbra), the University Hospital Center of São João (Centro Hospitalar Universitário de São João), the University Hospital Center of Central Lisbon (Centro Hospitalar Universitário de Lisboa Central), the University Hospital Center of North Lisbon (Centro Hospitalar Universitário Lisboa Norte), and the Hospital of Braga.

The interviewees were three males and two females, aged between 43 and 64 years.

Instrument

A script of the interview was prepared, and the first version of the script was submitted to an exploratory pre-test with experts in the field. For this pre-test validation, the methodology used was cognitive interviews, involving a panel of five experts with equivalent functions: service directors of other specialties from the Hospital Center of São João (Centro Hospitalar de São João), University Hospital Center of Porto (Centro Hospitalar Universitário do Porto), and Matosinhos Local Health Unit (Unidade Local de Saúde de Matosinhos).

The analysis of the cognitive interviews allowed the organizing of a set of guidelines in the reformulation of the script. The experts assessed the issues according to their suitability for the target population or cultural context, the objective formulation of the items, consistency of certain words or phrases, the degree of understanding, potential redundancies, among other aspects. As a result of this methodological procedure, some changes have been made in the formulation of words and expressions, as well as in the presentation format of some questions. The final interview script is detailed in Table 1.

In the semi-structured interviews, questions were asked about the work experience in the genetics service, more specifically (a) the general functioning of the service, (b) the patient care, (c) quality control, (d) challenges to health management, and (e) subjective assessment of opportunities for monitoring and improving health care in the area of genetics.

Data collection and analysis procedures

Initially, a registered letter was sent to all the directors of the country's genetics service, explaining the objectives of the study. Subsequently, an email was sent to confirm their availability. When conducting the interviews, the service directors gave their informed consent regarding participation in the study and authorization to audio record the interviews. In the cases of hospitals, we were not able to visit; we conducted the interviews remotely by videoconference on the Zoom platform.

The interviews were audio recorded with the consent of the participants, fully transcribed, and checked for its accuracy. The only person who accessed the content was the lead investigator of the study, ensuring the privacy and confidentiality of the data.

The total duration of the interviews was 8 h and 24 min, the average duration being 1 h and 41 min, the shortest interview duration of 1 h and 5 min, and the longest 1 h and 57 min. The interviewed service directors represent hospitals from the north, center, and south regions of the country.

Data collection was based on interviews with a sample of genetics service directors of the country; this being the only inclusion criterion. Although the script of the interview covered several topics, no coding category was previously defined, and the themes emerged from the data. The transcripts of the semi-structured interviews were read repeatedly and analyzed using the Braun and Clarke thematic analysis method (2006). This method is widely used to identify patterns and themes within the collected data, being appropriate when the research issue is broad and one intends to explore the topic in depth.

The data analysis process involved the identification of themes and subthemes within each interview, and later, the comparison of all interviews, the improvement of the subthemes, and the construction of a conceptual map were made.

Results

According to the methodological assumptions used in this article and the objectives defined for the study, thematic analysis of the transcriptions of the interviews resulted in four main conceptual themes: (1) specialty and technical developments, (2) structural difficulties, (3) potentialities, and (4) future directions. Figure 1 illustrates the structure of these conceptual categories, including subthemes. The categories emerged from the sequence of the various questions launched by the researcher in the interviews.

The context of genetics services emerged as a thematic block, which we do not consider relevant as qualitative data, but we consider it relevant for the better contextualization of the conceptual themes we present below. This block resulted from some issues that we consider relevant to perceive the historical and current framework of the performance of genetics services.

Table 1 Questions of the semi-structured interview used for contact with the directors

Questions
General service functioning
1. Can you briefly share the history of your service, also addressing the mission, vision, and values that support it?
2. In terms of organization and management, how is your service structured?
3. What are the main functions and activities that your service develops?
4. How many medical specialists and how many interns work in your service?
5. What is the workload of the doctors in your service?
6. What is the annual care activity they provide in the number of patients?
7. How often is the production evaluation done?
8. Is your service part of the National Health Service (Serviço Nacional de Saúde) referral network? How is this referencing done?
Assistance of patients
9. Can you briefly describe the conditions of access of patients to your service (self-referencing, medical referencing, teleconsultation, physical accessibility, others)?
10. What are the main reasons and typologies of consultations/patients?
11. Is there a mechanism for clinical screening of requests?
12. Is there a clinical registration methodology? How is it done?
13. Can you indicate the average waiting time for a consultation and the average waiting time for the diagnosis?
14. What is the response rate for the number of participants who has the result of the genetic test in the expected time of consultation?
Quality control
15. What kind of routine do you have regarding service meetings, clinical case discussions, and bibliographic review meetings?
16. Is there a culture of clinical quality assessment and auditing in your service? In what way are they carried out?
17. What are the quality assessment procedures or routines used in the management of your service?
18. What are the quality indicators used? How are these quality indicators monitored?
19. What is the access to funded actions of professional valorization and continuing training?
20. What role does research play in the organization and functioning of the service?
21. What methods are stipulated in the dissemination of good practices and clinical efficiency (guidelines, protocols of action, handling of complaints, others), and which ones are the most valued?
Challenges to health management
22. What is the role of the clinical expertise in genetics decisions (the professional capability of choosing the adequate genetic tests taking into account the clinical observation of patient's dysmorphologies)?
23. What is the role of genetics in the classification of diseases and what impact can it have on hospital costs?
24. Is there any mechanism to avoid unnecessary or redundant testing costs?
Subjective evaluation
25. Generally speaking, what are the positive aspects of how your service works?
26. On the other hand, what are the aspects that you think should be improved in your service?
27. What are the challenges you recognize to be the main ones in the day-to-day functioning of your service?
28. Do you foresee any future opportunities for improvement in your service?
29. What distinguishes your service from other hospital services? How do you think the genetics service is seen by the rest of the services?

The results are accompanied by the summary of the interviews and some excerpts from the transcripts.

Context of genetics services

According to the interviews, the description of the context of genetics services emerged as a thematic block. Thus, genetics services have as their main mission: provide quality medical genetics care to the population—genetic counseling, etiological identification genetics, prenatal diagnosis, pre-implantation diagnosis, integrate pre-graduate and post-graduate medical education, provide clinical advice in the field of genetics—discussion of cases and participation in service meetings of other specialties—and consulting of Complementary Means of Diagnosis and Therapy. In addition, they establish close collaboration with academic and university centers, as well as with other hospital services, national institutions, and primary health care. They seek to optimize the accessibility and equity of hospital care, also ensuring participation in reference centers, European networks and research projects, clinical trials, and dissemination of results in publications and scientific meetings.

The values of the genetics services mentioned by the directors are based on the following: (1) providing health care in a fair, high quality, and appropriate manner to the values of patients, (2) to privilege transparency towards patients and employees, (3) to produce cost-conscious activity, and (4) hold the team accountable for rights, duties, laws, and international or national guidelines.

Services have different organizational structures. The genetics service of the University Hospital Center of São João (Centro Hospitalar Universitário de São João) is governed by the Rector of University of Porto (Universidade do Porto) and the Administrative Council. On the part of

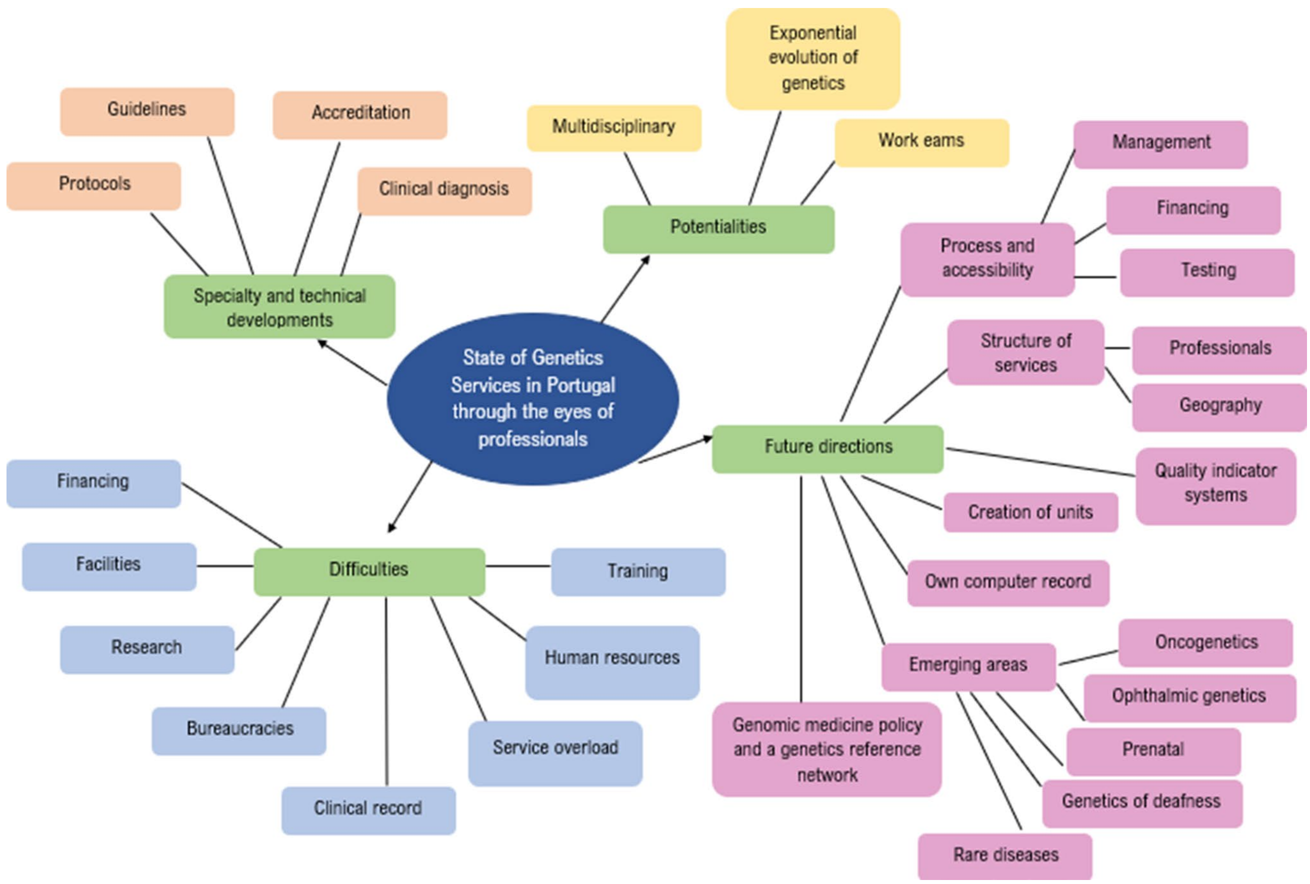

Fig. 1 Dimensions and subdimensions resulting from the analysis of the results

the University, it belongs to the pathology department. On the part of the hospital, it belongs to the Medicine Autonomous Management Unit (Unidade Autónoma de Gestão de Medicina). It is staffed by the service director, intern/residency doctors, medical specialists, graduated medical specialists, the secretariat of the faculty, and the various secretariats of the external consultation spaces. The genetics service of the Hospital of Braga is made up of doctors, secretariats of the consultation floors and the patient admission floor, the central archive, and the psychologist that supports the consultation, but it is part of an independent service. The genetics service of the University Hospital Center of Coimbra is integrated in the department of pediatrics and is made up of medical specialists, intern/residency doctors, laboratory technicians, a technical assistant, a nurse, and a psychologist. The genetics service of the Hospital Center of Central Lisbon is made up of medical specialists, intern/residency doctors, two administrative assistants, two nurses, and a psychologist that provides consultations despite not being part of the service. Finally, the genetics service of the Hospital Center of North Lisbon is made up of doctors responsible for clinical triage, tutors, a technical and laboratory coordinator, an administrative, and a psychologist. The Administrative Council is made up of the clinical director, the department of pediatrics director, the director of genetics, and, finally, doctors and other technicians.

As far as their composition is concerned, the genetics services interviewed are made up of 26 specialist doctors in medical genetics full-time, 5 medical specialists part-time, and 28 intern/residency doctors. Regarding the volume of consultations in 2019, ranged from 301 to 3134 first consultations and 283 and 7319 subsequent consultations, the average value is 2318 first consultations and 2983 subsequent consultations. All services reported that in 2020 there was a slight increase in the total number of consultations.

Specialty and technical developments

One of the themes that emerged from the reading of the data collected from the interviews addresses issues related to the evolution of services in recent years. One of the most mentioned topics by service directors was advances in clinical diagnosis, because of the appearance of more comprehensive panels and examinations.

The availability and use of genetic testing had and will continue to have an exponential increase. (P1)
The role of the clinic is always fundamental at various levels, however, over time it has been slightly fading in the sense that previously the study was very gene by gene, disease by disease oriented. Now the tests are more comprehensive and we often switch from genotype to clinic and from clinic to genotype. (P2)
More and more, we are able to propose studies having broader nature that go a little beyond the need to specify diagnostic hypotheses and that often give us results that we would never have been able to achieve through our observations. (P5)

Another topic refers to the greater and more detailed creation of guidance protocols for pathologies, clinical protocols and guidelines, patient guidance, and standards for referral of patients. They also mention the accreditation process as fundamental to the growth and credibility of genetics services.

We have protocols, we've been having in collaboration with the other services. It's something that is done little by little. Just the day before yesterday I had a service meeting where the protocol for study was discussed, for prenatal genetic evaluation. (P3)
We have guidance protocols for certain pathologies, we have guidance protocols for the use of complementary diagnostic means, that is, genetic testing in this case. (P5)
There are some audits that occur within the framework of the department's accreditation process. Therefore, the department was accredited by AXA, a model of the DGS, in 2019. (P4)

Structural difficulties

Another emerging theme of the interviewees' discourse concerns the structural difficulties in the current functioning of genetics services. The following subthemes are evidenced in this theme: financing, facilities, research, human resources, clinical record, service overload, bureaucracies, and training.

Based on the unanimity of the interviews, the lack of funding for training and research, the lack of genetics literacy of professionals from other specialties and in particular from general and family medicine doctors is highlighted, which often leads to misfit test requisitions.

One way to combat monotony is to have problems to solve in the laboratory related to the thing that causes us monotony. Now there's something there related to the previous question, isn't there? Who pays? Who's funding it? (P3)
We are allowed to attend training while keeping our monthly salary, but there is no budget for subsidizing the travel or the application in the training activities. (P1)
So, a time for training is conceived. There is no support for its completion. Therefore, all of this is done at our own expense in a specialty where support, particularly from pharmaceutical companies or whatever it is, is almost none. So, in fact, people do a lot of these things or try to do, but absolutely at their own expense. Our own training center, the training that is done has nothing to do with what is needed for our technical area, right? (P4)
And, on the other hand, one area where we cannot be very successful is improving the competence of other specialties in the handling of genetic testing. When I say handling I mean to ask, to interpret the result and to guide the patient and the family. (P1)
Compulsorily associated with the training of family doctors, especially family doctors that are the direct faces with whom these users contact. (P5)

Other difficulties pointed out relate to the overload of services due to the number of patients, the lack of professionals, the pressure from the higher bodies for the results, and the waiting list for consultation being out of control. Lack of professionals does not mean lack of doctors only, but the absence of support professionals, whether it be nurses, psychologists, or administrative assistants.

Things to improve would be having a larger medical team, also having a more numerous team with non-medical elements and having a work environment that is more facilitating of complementary activities to the direct provision of supportive care. (P1)
I said that one of the missions of the service is to create conditions for people to have a satisfactory career during their professional path [.] and this should be more integrated into the work plan of everyone, but no. In practice this quality index talking about the number of consultations, that's what counts. (P4)
We have nowhere to turn, we have nowhere to make consultations. We have many things to do, therefore, I think the main challenge at the moment is the lack of administrative resources and the lack of material resources. (P4)

In addition, the directors reported failures in the service facilities, in some cases due to the physical space being maladjusted to the number of professionals, in others due to inadequate location within the hospital center.

There's no room! There is no room, which means, at the moment, for 2022 I warned the Ministry of Health that, despite having formative capacity, I have no logistical capacity. So, we have an important space restriction at this point. I don't like to say restriction

because we weren't restricted at all... we have space constraint and we have constraint of secretariat support. These are our main problems at the moment. (P3)
And we are obliged, for lack of offices to distribute our schedule at proper hours, to make some consultations in a room without any conditions. So, it also happens to us. Many of our adult consultations are made in obstetrics offices, so these are places with examination tables with legrests and deficient things like this. The ones we do here at the service, for example those of predictive, are in a place that is a small collection room, there is no window, it's tiny. So, I'd say there aren't great conditions. (P4)
I'm a little sorry that a service that should be one of the quality brands of a hospital center is like this, and I can tell you that that's my position… When we enter this service, first we go through the storage, we go through the uniform and such. And it's sad that the places and the people who work in them are not given the relevance they deserve. And my team deserves to be recognized for its merit and its value! (P5)

Finally, they denounced the huge bureaucracy required in all hospital procedures and the fact that investigation is often left in the background, most of the time, for lack of time, lack of own projects, and with the participation being reduced to only phase III clinical trials.

The investigation part is practically all done outside the Hospital Center to be able to get something done because the amount of papers and authorizations that are needed limits everyone and causes anyone to give up. Let's hope that changes in the near future! (P5)
Investigation is the poor relative. The interns, fortunately, really have to do things and really have to write things and, therefore, they do and they write, but this is ripped from the extra hours of everyone: of the interns, of the tutors, of my own. No one can do this in their forty hours of work. (P4)
Also now, 2021, research activity is just the participation in phase III clinical trials. (P1)

Potentialities

Genetics is a very recent and unexplored health area which increased and evolved in latest decades due to technological advances. Thus, this section focuses not only on genetics current work but also on growth potential this area can achieve. In this regard, understanding what has been done with this aim can help draw lines for further research and work.

Thus, service directors argue that it has been essential to set up more cohesive work teams, multidisciplinary, constantly in conjunction with other specialties.

They refer across the board the importance of meetings with other experts, the need to improve the skills of other specialties in the handling of genetic tests, and their fundamental presence in group consultations.

So, the different specialties in the field of pathology diagnosis of the respective areas are increasingly asking for genetic studies, that is absolutely true. It is also true that this is pertinent. On the other hand, the best studies for the nominations are often not requested and, therefore, a very large national work would have to be done here. (P4)
The opportunities for improvement is to give greater support, when we can balance human resources with what is demanded of us. That is, multidisciplinary consultations with others, consultations and multidisciplinary meetings with other services in which we'll probably now start with oncology risk consultation and the collaboration. We already have the prenatal diagnostic consultation, but there are other consultations that may arise and that would be useful. (P2)

Future directions

On another emerging dimension from the interviews, the future of genetics is debated. The process and accessibility are included, where the experts interviewed assume the importance of management, financing, and testing. At the level of management, they defend that it is relevant to manage in the best way the waiting list of patients and the expectations of patients and professionals, to end the investigation that happens outside of working hours, to dematerialize genetic testing requisitions, and to give greater management power to intermediate management bodies.

At the moment, it is characterized by having a very large waiting list and the patients are expecting that 'when it comes, it will all be solved' or they have no hope anymore and have gone elsewhere, or are tired of the waiting. (P2)
The other is to end, for real, the 3F's regime [research and training on holidays, days off and weekends]. I think this is absolutely important and it is absolutely important that the institutions integrate this. (P3)
We are currently at a very advanced stage in the dematerialization of genetic testing requisitions. They are still made on paper, but we are currently testing a beta version, the computer scientists call it that. It is a program that will allow to make an emission, to make an electronic requisition. So, this will be a challenge because an electronic requisition will raise the screening challenge to the service. (P3)
So, we needed more power for intermediate management, we needed a management that valued merit and

competence more and therefore had a human resources policy much more attentive to the quality of what is done. That had a human resources policy that doesn't exist. (P4)

My service can improve in everything, right? In the conditions it offers for the learning of the interns, in the ability to give differentiation opportunities to specialists in areas for which they are especially motivated, or in lightening the great pressure that we all feel at the moment to respond to the medical needs of patients or family members. (P1)

I wish the problem of genetic testing requests was simplified. We would save everybody so much money, we would make our lives so much easier. It's a very old struggle and it has to be solved. (…) I would like to have the ability, the possibility to have people working with more time and more people to take all my dreams and my projects to get a great service. A great service in the sense of having a service that could give all the answers to more people and in a shorter time. (P5)

In terms of investment, they consider the challenge of increasing health costs in recent years, due to the development of genetics and the emergence of generalized tests.

Genetic confirmation makes it possible to optimize therapy, has a big impact or can have on life expectancy and quality of life, and I may be mistaken, but I think this is done with an increase in health costs. (P1)

At the level of testing, they state that there should happen a simplification of genetic testing requests and that the tests are increasingly comprehensive, accessible, differentiated, and, therefore, should be cheaper rather than an exploitation by the pharmaceutical industry.

Things are expensive because there are other values that have nothing to do with medicine, neither with necessity, nor with truth. (…) And so, when I'm asked what is possible to do with genetics, you can do anything because genetic testing has an obligation to be almost 100 times cheaper. (P5)

On the other hand, service directors warn of the need for a change in the structure of services, not only geographically for medical support to reach the whole country, but also at the level of health professionals. In this topic, they warn of the need for more professionals and better qualified, for the differentiation of genetic experts, and for greater investment in the training of intern/residency doctors, general medicine doctors, family doctors, and doctors from other specialties.

At the moment, at least looking at my service, of course we need more people, but maybe before having more doctors, I need a set of other professionals that would come to boost much more the work of my doctors. (P4)

Genetics will increasingly be more and more important, and I think it will be - no one has any doubt that it will be - the science, the discipline, the specialty of the next century. About this I have no doubts! Now, the more relevance it has, the more responsibility it will have associated and clinical geneticists will increasingly have to know this and know how to move around in the various areas where they will be needed. This will imply that people have a very large training in various areas and be very well - the right word is structured – to have well-structured heads. This will pose a major challenge both in training and in the orientation of the interns. (P5)

So, more and more, patients arrive at genetics services already with the studies. (…) On the other hand, it's not just for us to realize what our role is and what we have to do, it's that in this exercise there would be a big gain in terms of cost management, right? Because this way we're clearly wasting money. That's for sure! (P4)

In addition, the relevance of the creation of units dividing patients according to the genetic conditions or subspecialties of genetics is addressed, creating risk consultations for example. In this sense, some of the emerging areas in the field of genetics and of special interest referred are: oncogenetics, ophthalmic genetics, prenatal, genetics of deafness, and rare diseases.

The basic idea will be to create units within the service itself, due not only to the number of patients we have, but also due to the pathologies we face and mainly due to the amount, the number of pathologies and users for certain areas that will have to be necessarily differentiated. (P5)

Finally, experts advocate the creation of their own informatic registry to address the various annotation gaps and data confidentiality that current hospital computer systems have and warn of the need for the application of quality indicator systems—often mistaken for production indicators.

My staff also writes in the S. Clinic all the information that is considered useful to the rest of the team that is following the patient, but that never exceeds the information that is contained, and it is obviously mandatory to ensure confidentiality in genetic terms. (P5)

The S. Clinic allows you to write confidential things. What is the problem with confidential information? It's just accessible to the doctor who wrote it. This is a problem that the S. Clinic has to solve because what the law says is that there is some information that should be sited on the genetic processes, accessible by genetics, it doesn't say it should be by the genetics doctor. (P3)

We can be with great numbers on the other side. This is me letting off steam, but what I mean is that quality indicators of these are indicators of production! So they divert what is the sense of quality, honestly, but we have to, of course we have to, because every month I'm bombarded with numbers with red dots and green dots. The point is to take the green dots out. (P4)

Nonetheless, they refer that a true genomic medicine policy and a genetics reference network are the key opportunity for services to develop, by redefining the role of genetics services in the context of Portuguese hospitals.

This had to do, on the one hand, in fact with the referencing networks in genetics, but on the other hand with the redefinition of the role of genetics services in the context of hospitals and the definition of how genomic medicine was organized in Portugal, right? All of these things that are yet to be done. While they are yet to be done, everyone asks for what they feel like... (P4)

Discussion

In this work, we explored the context of genetics services in Portugal to obtain a real perception of the current conditions of the practice of medical genetics. Considering the results obtained in the interviews and what is described by Epstein (2006), genetics services have historically been allocated to tertiary care, generally in large academic centers due to their level of expertise. This results not only in geographical and financial barriers to access, but also in psychological barriers (Hawkins & Hayden 2011).

According to the literature, advances in genetics research in recent decades to meet the growing demand from consumers (Unim et al. 2019) have had a strong impact on the health system with serious gaps in the provision of genetics services (Calnan et al. 2006). These gaps are created by disparities in the distribution of the workforce and limited funding (Chou et al. 2009; Roberts et al. 2014; Senier et al. 2015; Maiese et al. 2019).

With the evolution of technology, the availability of genetic testing and panels increases (Cornel & van El 2017), and the effects of this trend are long waiting times for consultations, increased potential for misinterpretation of genetic test results, and overworked professionals (Kaye et al. 2020). As genetic testing becomes more common, cheaper, and more useful, professionals have a greater need for genetic counseling in the area of primary care (Stoll et al. 2018).

New models of the provision of genetic services are explained in the literature and, converging with the interviewees' discourse, suggest the integration of genetics in all medical specialties (Korf 2002), the collaboration between different health professionals, the integration of genetics into primary care (Emery 2001; Starfield et al. 2005), and the redistribution of professional roles (Battista et al. 2012; Stoll et al. 2018; Unim et al. 2019).

The experts interviewed report that users are assisted by a wide range of health professionals who deal with specific aspects, but who do not always have an overview of the impact of genetic disease on the user and his family (Unim et al. 2017). They suggest, and studies indicate that training and education in genetics should be implemented by direct supervision, encouraging collaboration among professionals (Unim et al. 2020). The geneticist doctor then plays an important role in communicating with specialists and coordinating care for the whole family, avoiding the repetition of diagnostic tests and efficiently managing the available resources (Tizzano-Ferrari 2017). In this way, the activity of the geneticist doctor is increasingly involved in transversal fields with other doctors from different areas, laboratory, bioinformatics (Cassiman 2010), biostatistics, politics, and health education (Roberts et al. 2014). Professional roles developed more recently, such as genetic advisors and genetic nurses, have also been identified in a number of environments where they are important to support geneticist doctors in multidisciplinary teams (Unim et al. 2017). Certification of non-medical personnel trained in genetics should be mandatory (Unim et al. 2020).

On the other hand, several questions were raised in the results of this study on the integration of genetics in the context of primary care (Slomp et al. 2022). This challenge is particularly relevant in rural and remote areas, where the lack of genetics services is widespread, resulting in inequalities in access to health care (Hawkins et al. 2011). Thus, general and family medicine specialists are the first medical point of contact within the health system and the source of referral for most patients with genetic concerns (Battista et al. 2012). Literature indicates that the integration of genetics in primary care will allow care to be provided more fairly and with a more egalitarian geographical distribution (Hawkins et al. 2011). Appropriate referrals and timely diagnostics would thus be ensured (Paneque et al. 2016; Maiese et al. 2019).

Studies report that barriers to the effective provision of genetics in primary care, such as inadequate knowledge about basic genetics and inadequate referral to genetics, lack of detailed family history, lack of test supervision, poor doctor self-confidence, and lack of reference guidelines (Unim et al. 2020; Paneque et al. 2016; Mikat-Stevens et al. 2015) need to be overcome. As suggested by the experts interviewed and according to the results of several studies, some of the strategies can be greater education and training, the transmission of clear and updated guidelines on genetic testing, opportunities to discuss issues with a genetics expert (Unim et al. 2017; Cusack et al. 2021), and the involvement of doctors in genetic care and multidisciplinary consultations (Chou et al. 2021).

Another emerging theme in the interviews is the scarcity of professionals. Internationally, few countries are close to achieving the UK's Royal College of Physicians recommendation of a ratio of 6–12 genetics professionals per million inhabitants (Abacan et al. 2018). To mitigate the effects, long-term investments are needed to encourage students to enter the genetics profession. The genetics' professional capacity within the health system will need to be substantially expanded, and waiting times for consultations should be reduced (Maiese et al. 2019). The experts interviewed report that certain tasks that traditionally belong to the geneticist doctor, such as reviewing records and preparing for the clinic, can be done by an assistant. The hiring of these professionals will reduce disparities, respond to the growing demand for services (Chou et al. 2021), and ensure that the doctor can work on tasks that are more in line with their training (Stoll et al. 2018).

On the other hand, it has been reported in the interviews that the introduction of genetic counselors in the services can bring benefits to the practice, since the skills of the geneticist doctor and genetic counselor complement each other. The geneticist doctor's tasks include physical examination and treatments that are not part of the genetic counselor's practice (Madlensky et al. 2017). In contrast, the genetic counselor's tasks have a greater emphasis on user education and counseling skills (Doyle et al. 2016). Studies show that genetic counseling can lead to increases in knowledge, user satisfaction, perceived personal control, positive health behaviors, and perceived risk accuracy. Anxiety and conflicts of interest usually decrease after genetic counseling (Madlensky et al. 2017).

The experts interviewed indicate that policies aimed at graduate and postgraduate training in medical genetics are of paramount importance to improve the knowledge and competence of the various health professionals in the provision of genetics services. The adoption of standards of practice can harmonize the differences between countries in the education and practice of genetics (Paneque et al. 2016) and allow for the proper regulation of quality and competence in the provision of genetics services (Unim et al. 2020).

A repositioning of genetics in the culture of care for the user needs to happen, involving the reconfiguration of responsibilities of specialists in general and family medicine, the management of expectations of health professionals and users, the involvement of health administration, and the authentic recognition of the benefits and limitations of genetics in medicine (Chou et al. 2021). Nevertheless, it is necessary to increase the number of specialists in medical genetics, recognize the specialty of laboratory clinical genetics, and invest in certified genetic counseling programs for health care professionals (Encina et al. 2019). Thus, the success of genetics services will require an organization along primary care, other specialties, and public health (Harris et al. 2000).

Another topic addressed by the interviewees is the lack of data from the service on quality and satisfaction. Genetic data gaps are systemic, as currently there is no robust collection of data about the access to genetic services (Kaye et al. 2020). Because of this, establishing a national network of genetics services is essential to fulfill this and to take advantage of the existing public telemedicine (Encina et al. 2019). Telemedicine becomes increasingly useful to improve access systems and the quality of genetic follow-up delivery to distant users and families (Kaye et al. 2020; Kubendra et al. 2017).

According to the literature, facilitating the acquisition of medicines and medical devices for rare diseases is an important step in the role of genetics. It is essential to encourage price negotiation and competition in the pharmaceutical industry (Encina et al. 2019). In that regard, it is essential to encourage EU member states to develop national rare disease plans in their health policies to ensure equal access to prevention, diagnosis, treatment, and rehabilitation (Battista et al. 2012). Such international support and cooperation should ensure that common policy guidelines are developed, and knowledge is shared in specific areas, including research, centers of expertise, access to information, orphan drugs, and screening (Battista et al. 2012; Encina et al. 2019). In this sense, the promotion of international strategic partnerships is a key factor for environments with limited resources, as it is the case of our country. Collaborations with well-funded international research groups create benefits for all parties involved through training, knowledge transfer, and participation in multicentric projects (Encina et al. 2019).

Other studies indicate that the creation of a free public information portal for genetic diseases should be carried out with the support of the Ministry of Health to improve the knowledge of the population (Encina et al. 2019). Collaboration between government, genetics services, and universities is necessary to identify priorities in research funding (Unim et al. 2017). The ability to translate complicated findings to individuals who lack advanced health literacy skills is extremely pertinent (Roberts et al. 2014), so that they are able to make autonomous decisions about their health care (Hawkins et al. 2011).

Finally, several studies highlight the role of associations of people with disease in the debate about equity and social protection and in the intervention in public policies (Encina et al. 2019). Considering the role of these associations, often the first resource of people with genetic diseases, their social value in this context of restructuring can be immeasurable (Costa et al. 2022). Associations of people with disease strengthen cohesion and local work by raising awareness, supporting education, and participation in research and clinical trials (Encina et al. 2019).

Limitations

This study may have some limitations. It is essential to highlight the convenience sample that only represents a part of the reality of the professionals of the country's genetics services. It is worth noting, however, that the directors of the main genetics services were included, as representatives of their clinicians and with motivation to participate in this process. The lack of a public, complete, and current list of health entities that provide genetic consultations makes the sampling process difficult.

Nevertheless, this work has an emerging character, given the reality of hospital genetics in Portugal. Although some caution is needed in the analysis of the results due to the limitations mentioned, the study is particularly relevant because it underlines pertinent information about the current state and needs of genetic services, reinforcing the imminent lack of structural change.

Conclusion

Genetics, regarded as the specialty of the future, currently plays a prominent role in the well-known post-genomics era. In Portugal, medical genetics needs to redesign its place in the health system.

There is an urgent need to increase the number of medical geneticists, genetic counselors and other health care professionals, well integrate at genetic services. There is also a need to improve genetics education and training in the primary health care and in other medical specialties. The importance of funding for training and research and the relevance of time available for these tasks emerged from this study. Constraints are reported in the physical facilities of the services as well as pressure from the higher hierarchies for specific outcomes, and the need to simplify procedures for genetic testing request. In addition, the creation of special units within the services acconrding to the specific diseases or area is suggested, as well as the creation of proper informatic registry to fulfill the current gaps.

A real policy for implementing genomic medicine is necessary. It should include the redefinition of the role of genetic services in Portuguese hospitals and the entire national health system, as well as a better organization and composition of services in a multidisciplinary and user-centered approach.

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Author contribution CC: study design, data collection and interpretation, writing of the manuscript.

MSL: critical review of the work and approval of the final version of the manuscript.

LFA: critical review of the work and approval of the final version of the manuscript.

MP: data collection and interpretation, critical review of the work, and approval of the final version of the manuscript.

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Declarations

Competing interests The authors declare no competing interests.

Conflict of interest The authors declare no competing interests.

Protection of people The authors declare that the procedures followed were in accordance with the regulations established by the heads of the Clinical And Ethical Investigation Commission (Comissão de Investigação Clínica e Ética) and in accordance with the World Medical Association Declaration of Helsinki updated in 2013. Informed consent was obtained from all participants to be included in the study.

Data confidentiality The authors declare they followed the protocols of their work center on data publication.

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