



Genetic counselling supervision: Luxury or necessity? A qualitative study with genetic healthcare professionals in Portugal

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ABSTRACT

In recent years, there has been a significant technological evolution in the field of genetics, leading to an increase in the number of professionals working in medical genetics and, consequently, a tremendous growth in genetic counselling. At the same time, there has been a growing recognition of the parameters on which to base a safe practice, not only regarding the technical skills of the professional but also regarding their counselling skills, including relational and empathy skills and the acknowledgement of the emotional impact that genetic counselling practice can have. However, despite this growing knowledge, there are still significant differences between the various European countries, and one area where this discrepancy is particularly evident is genetic counselling supervision. Thus, if there are countries where genetic counselling supervision is not even known by the professionals, there are others where it is mandatory for practice. This research had as an objective to understand if and how genetic counselling supervision is provided in Portugal, to identify routines, challenges and impacts of genetic counselling that should be explored in a supervision process and comprehend how professionals believe supervision should be conducted to be effective. A total of sixteen medical geneticists from main Portuguese genetic services were present in two online focus groups. None of the participants had access to genetic counselling supervision as a programmed routine and there was a consensus that a service of this kind would be particularly important for the professionals as genetic counselling has frequently challenging and emotional moments. Aspects regarding clinical supervision, the characteristics of the supervisor and the practical aspects of genetic counselling supervision implementation were also mentioned during the discussions. These results highlight the relevancy of the establishment of GC supervision routines and standardized guidelines in our country, as well as a need for evidence-based research focused on its impact at professional and practice level.

1. Introduction

In Portugal, as in many other European countries, the field of genetic counselling has emerged as an independent scientific discipline relatively recently (Paneque et al., 2015) - Medical Genetics was officially recognized as a medical specialty in 1999, and it gained recognition from the European Union in March 2011 (Costa et al., 2022). Despite these significant milestones, the formal recognition of the genetic

counselling profession in Portugal remains an ongoing challenge (Costa et al., 2022) and the role of the genetic counsellors has yet to be formally acknowledged and regulated within Portugal's healthcare system.

The inception of the first master's degree program in genetic counselling in Portugal occurred in 2010. This marked a pivotal moment in the efforts to advance genetic counselling services and education.

Genetic Services in Portugal are integrated into the National Health System (SNS), meaning that all professional salaries, consultations, and

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genetic tests are financed through public funding (Costa et al., 2022). Genetic services are currently not integrated into primary healthcare settings, but rather they are offered at larger central hospitals throughout the country. These services include genetic counselling, which is conducted by medical geneticists. Typically, referrals for genetic consultations are initiated by primary care providers or by colleagues from various medical specialties (Costa et al., 2022).

Studies conducted in Portugal over the last few years have identified several difficulties experienced by professionals working in genetics (Paneque et al., 2012, 2014, 2015; Guimaraes et al., 2013; Costa et al., 2022). The challenges encompass several key aspects: (1) an overwhelming demand for services due to a high volume of patients, (2) a shortage of healthcare professionals, (3) pressure from higher authorities for quick results, (4) backlog of consultation requests - the scarcity of professionals encompasses not only doctors but also the absence of supporting staff, including nurses, psychologists, and administrative assistants, (5) insufficient funding for training and research, coupled with a lack of genetics literacy among professionals from various specialties, (6) inadequacies in physical facilities, either due to inadequate size or suboptimal locations within hospitals, (7) bureaucratic hurdles in hospital procedures (Paneque et al., 2012, 2014, 2015; Guimaraes et al., 2013; Costa et al., 2022). Other difficulties pointed out are generic and concern the organization of the services themselves, the articulation with the Portuguese genetic network and the lack of harmonization for practice and of specific tools and routines for quality assessment (Paneque et al., 2014; Costa et al., 2022). These issues collectively impact patient access to genetic services, resulting in delayed care and strain on healthcare professionals, raising concerns about burnout and incomplete patient care.

On the other hand, when examining the global practice of genetic counselling (GC) in the world, we find that there are more than seventy different master's degrees, with different curricula (Middleton et al., 2007; Ingvaldstad et al., 2016), verifying an enormous variability of practices between countries and even within the same country. Until recently, only the United Kingdom, Portugal, Romania, France, and Spain had MSc education in Genetic Counselling (<https://www-esbg.org/408.0.html>) and, consequently, access to genetic counselling services is not consistently available throughout Europe (McAllister et al., 2016), and in some countries, it is conducted by professionals without specialized training (Skirton et al., 2010).

Genetic counselling supervision (GCS) is precisely one of the areas in which there is no structured guiding legislation common to distinct parts of the globe. Thus, if in some European countries, such as the UK, access to supervisory services is mandatory for the professional practice of genetic counselling, in other countries, like Portugal, there are no standards or guidelines in this regard (Middleton et al., 2007).

The focus in genetic counselling supervision is the emotional and psychosocial issues of the professional (Evans, 2006; Hawkins and Sholet, 2000). These issues may be related to ethical dilemmas, transference and countertransference and difficulties in dealing with specific situations that emerge throughout genetic counselling. Consequently, a crucial aspect of GCS involves providing professional support, and creating a space to reflect on one's own feelings and reactions in the different GC processes the professional accompanies (McCarthy et al., 2003).

However, despite being an emergent subtheme within the field of genetic counselling (GC), there is evidence of the relevance of genetic counselling supervision (GCS) (Rothwell et al., 2021). As evidence of its recent development, the first study on genetic counselling supervision emerged in 2002 (Hendrickson et al., 2002). The author concluded that there was a lack of guidelines upon which supervisors could base their efforts to improve the genetic counselling process. The study also underscored the importance of conducting further research in this area and the necessity of organizing specific workshops for professionals who would be engaged in genetic counselling supervision (Hendrickson et al., 2002).

Recent studies have identified specific areas that should be addressed during GCS. Research on the cultural competence of supervisors, concluded that one of the key areas of GCS should be raising awareness of multiculturalism and its implications (Paneque et al., 2015). Other authors focused on the extent to which professionals were aware of issues related to the ethical boundaries between consultant and professional and noting numerous violations of basic rules of conduct and ethics, suggesting that this should also be a focal point of genetic counselling supervision (Gu et al., 2011).

Thus, despite the limited literature on this topic, existing empirical data support the need for genetic counselling supervision as it offers a multitude of benefits within the field of genetic counselling: (1) it serves as a valuable platform for professional development, enabling genetic counsellors to continually refine their skills, enhance their knowledge base, and navigate intricate ethical dilemmas; (2) through regular supervision sessions, counsellors can effectively address challenging cases, develop robust communication strategies, and gain insights into the emotional and psychological aspects of patient care; (3) supervision fosters a supportive environment for personal growth and self-awareness, aiding counsellors in managing the emotional demands of their role; (4) ensure genetic counsellors remain competent and well-equipped to meet the evolving needs of their clients in the ever-advancing field of genetics and genomics (Middleton et al., 2007; Hawkins and Sholet, 2000; Rothwell et al., 2021; Hendrickson et al., 2002; Bernard and Goodyear, 2008).

Also, the European Board of Medical Genetics (EBMG), in the document "Counselling Supervision for Genetic Counsellors" (<https://www.ebmgeu>) recommends that genetic counselling professionals use both: clinical supervision and genetic counselling supervision. This document considers clinical supervision as involving discussion of cases within multidisciplinary teams and genetic counselling supervision as a way of exploring the emotional and psychosocial impact of genetic counselling on the professionals. In short, they consider that genetic counselling supervision will allow the professional: (1) to obtain psychological support, (2) to recognize how their personal characteristics can influence the work they develop with the consultants, (3) to explore related issues with transference and countertransference, (4) to support the professional to deal with all the emotional load of the work he develops, (5) to help the professional to perceive ways to improve his work, and (6) to reduce the probability of developing burnout.

Considering the specificities of genetic counselling, the emotional impact of GC and the shortcomings already mentioned earlier, this study, which focused on the Portuguese context, aimed to: (1) understand if and how genetic counselling practice is supervised in Portugal; (2) identify routines, challenges and impacts of the genetic counselling processes that should be explored in a supervision process; as well as (3) to identify factors relevant to effective supervision implementation.

2. Method

Focus groups were chosen as the methodological approach for this research because this method facilitates group interaction, capture collective views, provide natural and interactive discussions, explore sensitive topics, allowing the exploration of diverse perspectives and the generation of a rich data set^{27,28}. As this study aimed to gain a comprehensive understanding of participants' experiences and perspectives in GCS it offered a main advantage: through the group dynamics, participants could build on each other's ideas, share experiences, and engage in nuanced discussions, providing a deeper reflection of their understanding on this topic (Morgan, 1996; Stewart and Shamdasani, 2014).

2.1. Participants

Taking into consideration the study's objectives and the

characteristics of the Portuguese national healthcare system and of the genetic services, medical geneticists meeting the following criteria were invited to participate in this study.

- (1) Medical genetic specialist working in a genetics service in Portugal.
- (2) Member of the Board of the Specialty College of Medical Genetics.
- (3) Member of the Board of the Portuguese Society of Human Genetics.

As exclusion factors, we defined.

- (1) Genetic Counsellors (as the profession is not recognized in Portugal).
- (2) Medical specialty interns currently serving in a genetics service.

The distribution of participants into the two focus groups was based solely on the availability for the proposed dates and did not follow any specific criteria.

Five medical geneticists were present in the first focus group and eleven were present in the second one, in a total of sixteen participants, corresponding to 33% of medical geneticists conducting genetic counselling in Portugal.

These professionals practiced genetic counselling in the following Portuguese genetic services: University Hospital Center of Coimbra (Centro Hospitalar Universitário de Coimbra), University Hospital Center of São João (Centro Hospitalar Universitário de São João), University Hospital Center of Central Lisbon (Centro Hospitalar Universitário de Lisboa Central), University Hospital Center of North Lisbon (Centro Hospitalar Universitário Lisboa Norte), Hospital of Braga and Portuguese Institute of Oncology – Porto (IPO- Porto).

2.2. Procedures

An email was sent to all genetic service directors informing them about the study design and objectives. They were requested to provide their availability to participate in an online focus group and to identify other team members who met the requirements to participate. Two dates were established based on the participant's availability.

A script which included: (1) objectives; (2) expected duration of the discussion (2 h and 30 min); (3) questions to the participants (Table 1); (4) online tools to be used during the focus groups and (5) basic rules, was created, and used as a guideline to the researchers, ensuring that they understood their roles during the focus group, and that the participants were aware of the different moments that were prepared.

Table 1
Instrument.

1. Given the definition^a of genetic counselling supervision do you currently have access to any service of this nature? (Online survey).
2. Considering the presented definition of genetic counselling supervision, which routines do you use to share the impact or difficulties of emotionally difficult processes?
3. How can this type of emotionally impactful consultations affect the professionals?
4. Identify moments, challenges, impacts that would make sense to share in a supervision process (Mentimeter)
5. What form would supervision have to take to be successfully implemented:
 - (a) Among peers vs. with other professionals
 - (b) Within the same service vs. out of service
 - (c) Group supervision vs. individual supervision
 - (d) Financed by the institution and regulated as in other countries?

^a At the beginning of the focus groups the following definition of GCS was presented: 'Genetic Counselling Supervision (GCS) is the process by which genetic healthcare professionals, on a regular and structured basis, explore and share emotionally impactful moments, dilemmas, and situations that arise from the specificities of GC) and that can affect the quality of the service provided and the mental health of the professional'.

The day before the scheduled dates, an email was sent to remind participants of the focus group's timetable, the link to access the online meeting, and the script.

At the beginning of each session, the researchers introduced themselves, restated the study objectives, and reminded the participants of the following rules: (1) Provide informed consent to participate and be recorded, (2) Mute the microphone and enable it only when speaking, (3) Select "raise your hand" when wanting to contribute, (4) Respect the allocated time for each question, (5) Use the chat for technical support only, (6) Keep the video on to facilitate flow of conversation, (7) Everyone is invited to participate actively.

The focus groups were conducted by two researchers from the group: MP and LG. Both researchers are psychologists with specialized training in genetic counselling.

Privacy and confidentiality were mentioned, and verbal authorization was requested to record the session. Some of the participants were acquaintances in a professional context of the researchers, so various techniques were employed, including methods that facilitated anonymity, to ease disclosure. The established script was flexible and was adjusted as the discussion progressed.

2.3. Measures

The two online focus groups were conducted with prior alignment and previously developed questions (Table 1). As is customary in focus groups, the questions served as catalysts for discussion. To gather participants' opinions, we utilized various techniques tailored to the specific objectives of each question.

For question number 1, we used an online survey with yes or no answers. This direct approach allowed us to check if any of the participants had access to GCS and served as a basis for initiating the subsequent discussion. For question number 4, we utilized Mentimeter, an interactive online tool available through the Zoom platform. This question - *Identify moments, challenges, impacts that would make sense to share in a supervision process* - delved into individual experiences, identifying important moments, challenges, and impacts to share in GCS. Given the diverse composition of the groups, including heads of service, junior professionals, and experienced professionals, we recognized that the use of Mentimeter would promote transparency and encourage participants to focus on the themes they deemed significant for GCS. The answers were displayed anonymously on the screen as participants added comments, serving as a starting point for the ensuing discussion. The other three questions were discussed verbally. All the questions, including questions 1 and 4, sparked engaging discussions, interactions, and dialogues among the participants. The questions asked in both focus groups are present in Table 1.

2.4. Data analysis

All data obtained was recorded and transcribed verbatim.

With the objective of identifying emerging themes related to genetic counselling supervision in Portugal, data was analysed using thematic analysis (Maguire and Delahunt, 2017; Prokopia, 2023). With this approach we aimed to achieve an in-depth understanding of the challenges, perceptions, and needs of genetic counselling professionals regarding supervision.

The transcripts of the focus group discussions were analysed following the principles of thematic analysis (Maguire and Delahunt, 2017; Prokopia, 2023): Initially, two of the researchers (LG, RB) present in the focus groups read and re-read the data to familiarize themselves with the content and generate initial ideas. Individual conceptual diagrams were then created (Fig. 1 – presents the conceptual diagram of LG), highlighting key ideas and emerging concepts. These individual analyses were then shared with a third researcher (MP) that compared and contrasted the individual analysis. Through collaborative discussions, common themes were identified and refined. The emerging



Fig. 1. Conceptual diagram.

themes and subthemes were then defined based on the convergence of the researchers' analyses. When discrepancies arose, they were discussed within the group, with the researchers presenting their arguments. If differences persisted, the majority view was accepted. Additionally, unique themes that appeared less frequently or were mentioned only once in the participants' dialogue were brought forth for discussion and, when considered to have relevant importance to our theme, were also included in the results.

3. Results

Overall, there was a substantial consensus regarding the importance of supervision for fostering continuous improvement in professional practice, improving service delivery, and enhancing the mental well-being of professionals. There was also unanimity on the preferred approach to supervision, involving a blend of group and individual supervision led by an external supervisor. The most notable divergence arose from two participants who considered that supervision should be categorized under occupational health and not be obligatory. They argued that this need wasn't unique to genetic counselling, as other healthcare domains also deal with emotional aspects.

Given the richness of all the contributions, as well as the multitude of factors present in supervision, we will conduct a more detailed presentation of the results.

All the participants considered that they did not have access to

genetic counselling supervision as a programmed routine of support and reflection on their clinical practice.

Through thematic analysis of the verbatim transcriptions from the focus groups, two significant themes emerged (Table 2).

- (1) Unfamiliarity with the concept of genetic counselling supervision.
- (2) Supervision as a potential source of continuous professional development

3.1. Unfamiliarity with genetic counselling supervision concept

3.1.1. Strategies used to deal with stressful practice

During the focus groups, none of the participants had full knowledge of what a genetic counselling supervision process consisted of, revealing, however, that they felt the need for a regular routine and formal professional support, given the specific and challenging characteristics of genetic counselling:

"It is something that we had already talked about in my service, quite informally, and how important it could be for us to make this into something more structured and organized" (P2, FG1)

"This, without a doubt, should be something structured and included in the service work routine." (P1, FG1)

Table 2
Themes and subthemes obtained throughout the use of thematic analysis.

Theme 1 Unfamiliarity with the concept of genetic counselling supervision	<i>Strategies used to deal with stressful practice</i>	<ul style="list-style-type: none">- Informal routines- Case discussion in service meetings- Case discussion with the service psychologist- Clinical supervision of junior professionals
	<i>Unclear vision of how genetic counselling supervision should take place</i>	<ul style="list-style-type: none">- Never thought on this issue before -Variable, according to the professional characteristics- Combination of individual and group supervision- Supervisor with specific training in supervision and/or knowledge of genetic counselling- Occupational health affection- Regulated or non-regulated
Theme 2 Genetic counselling supervision as a potential source for continuous professional development	<i>Supervision as a way to develop specific skills</i>	<ul style="list-style-type: none">- Development of specific technical skills- Development of communication skills- Transmitting bad news- Managing personal characteristics and life history- Managing pressure from users and colleagues from other specialties- Avoiding crystallized errors- Improving service quality
	<i>Supervision as a way to protect the professional's mental health</i>	<ul style="list-style-type: none">- Preventing recourse to secondary systems – medication, exercise, alcohol- Preventing recourse to defence mechanisms/self-protection behaviours- Preventing isolation- Managing thoughts outside working hours- Emotional impact of medical activity

Due to the emotional and ethical implications of genetic counselling, several of the participants mentioned that they often resort to informal supervisory practices:

“(…) in fact, almost all of us have our routine, either having lunch, or taking breaks with our peers, and almost all of us know who is having challenging appointments and then we go to lunch with that person who ends up getting to know us.” (P1, FG1)

In addition to these informal moments, it is also referred that service meetings are used, usually, to discuss ethical dilemmas or situations that are having more emotional impact:

“The dilemmas are also taken to the service meetings, so that there can be some generalized opinion about them” (P1, FG1)

“In the service meeting, there is a space for one of you to discuss issues that may arise from more emotionally demanding situations” (P3, FG1)

The professionals also mentioned that they often turn to the service psychologist when they need to further explore a case or situation or when they feel that their own specific characteristics and their life history are not allowing them to perform more adequately in a given

situation:

“(…) Occasionally, and I do not know with what frequency, there is an informal intervention in this context, when a service element dialogue with the psychologist who performs genetic counselling technician functions in the service.” (P6, FG2)

3.1.2. Unclear vision of how genetic counselling supervision should take place

When questioned about how the genetic counselling supervision should take place, as well as what rules should guide it, the answers covered various spectrums, with no convergent conclusion.

There were several professionals who mentioned that genetic counselling supervision should be variable according to the professional characteristics:

“There must be different options because we are not all the same.” (P5, FG1)

Also, in relation to how genetic counselling supervision should occur, individually or in a group, or to the periodicity and characteristics of the supervisor, the answers were not conclusive:

“But I really agree, and I see myself in a group and in individual moments. I think we benefit a lot from discussing these issues in a group because we perceive our colleagues to have the same difficulties and we see ourselves in them and they in us and the strategies can be useful for everyone but there will be more particular situations in which we will feel more comfortable and we will be able to go to a deeper level of reflection and draw strategies if we do it individually, in a one-on-one conversation.” (P2, FG1)

“Someone who had a difference knowledge, like someone who had specific training in genetic counselling supervision, I think it could be an asset, even because being from the area they could recognize in us signs that we may not recognize and that are warning signs for something you mentioned earlier – a burnout situation or when we go beyond the limits of what is reasonable and acceptable” (P2, FG1)

“It requires this agenda time to think about how things are being done ... and then it necessitates the sharing of various experiences ... I think one learns ... I think ... a group with an outsider organizing the group.” (P4, FG1)

In relation to regulation, the responses were not convergent - part of the participants considered that genetic counselling supervision should be mandatory and regulated while two of the medical geneticists considered it should be optional:

“It should be individual, optional, outside the service and in the area of occupational medicine and occupational health that already has some specific activity more in some issues than in others, related to burnout, with this and with that and which should be in this area, evidently being an occupational health service of the institution, it would be reimbursed by the institution.” (P6, FG2)

3.2. Supervision as a source of continuous professional development

3.2.1. Supervision as a way to develop specific skills

Although it was frequently mentioned they had never thought about this topic before, professionals referred some of the areas in which they considered that supervision could be beneficial to their practice.

One of the key findings is related to the enhancement of specific skills required by professionals involved in genetic counselling. Communication skills and delivering difficult news were among the most discussed topics when answering question number 4 using Mentimeter (Table 3).

Also, the management of personal characteristics, as well as addressing situations related to the professional's personal history, were identified as areas in which GCS could be beneficial.

Table 3
Answers to question number 4, using Mentimeter.

Identify moments, challenges, impacts that would make sense to share in a supervision process				
Communication Challenges	Emotional Challenges	Mental health Issues	Ethical Issues	Continuous Professional Development
Transmission of bad news.	Immediate consequences. Reactions. Dealing with unexpected reactions.	Situations of despair, feeling blocked, desire to give up because of a consultation.	Managing professional opinion disagreements/Managing deep beliefs.	New ways of doing things. Not getting stuck in routines and habits. Evolving.
Difficulties/communication barriers.	Pressure from patients and/or colleagues.	Avoided listening to the patient to protect myself.	Dealing with deep disagreements with the consultant's decisions.	Predictive tests. Prenatal diagnosis
Situations with difficult communication processes.	Impact of the results on patients.	This "explore" can be variable, sometimes. We may feel the need to verbalize immediately, sometimes only later.	Genetic counselling in families that have a history similar to the doctor's life history.	Deal with lethal and/or degenerative diseases.
Situations in which there was not enough time in consultation to listen and dialogue with the family.	Excessive empathy.	Being able to abstract from situations in post-work hours./Situations that made you think about an appointment for a long time, without being able to deal with the matter.		

“Genetic counselling processes in situations close to family history”.
“(…) because I am a very empathetic person, sometimes the emotional impact of consultations, this type of consultation is huge (…)
Dealing with user pressure and peer pressure was also addressed:
“Pressure from patients.”
“Pressure from colleagues in other specialties.”
“Managing disagreements of professional opinions.”

The participants also verbalized that having such a service would be of extreme relevance to the enhancement of their skills, well-being, and a source of continuous improvement, and that it would be a luxury regarding the actual Portuguese panorama where structural changes are urgently needed.

(…) We are so dominated by all the other things that stopping to talk about a luxury is difficult, but it is important because we can evolve here”

Finally, the professionals considered that carrying out supervision could be an effective way of overcoming crystallized errors, reflecting on new ways to proceed and of continually improving their practice:

“Learning to deal with these emotions and developing strategies and tools that will allow us to make continuous improvement, we will also be able to play a better role for our patients at the very moment of the consultation, in daily clinical practice, and that will reflect in the quality of the service we provide and I think it will allow the people who are on the other side, in front of us, to leave feeling better (…) (P2, FG1)

“There are crystallized errors in which we no longer have the capacity for self-criticism, and it is obvious that it is not that sharing would help with that ...” (P4, FG1)

3.2.2. Supervision as a way to protect the professional's mental health
Issues related with the professional mental health also emerged, and some of the ways that are used to protect their mental health were explored.

Various professionals referred to use secondary systems to deal with the impact of genetic counselling consultations, namely physical exercise, food and even medication and alcohol:

“Substance abuse, be it chocolate or alcohol (…) medication, alcohol, drugs ... “(P4, FG1)
“(…)physical exercise is one of my best ways to manage problems I have” (P5, FG1)

Some professionals mentioned that they developed defence mechanisms that allow them to deal more positively with the impact of some of the genetic counselling processes:

“We all end up acquiring our own defence mechanisms and experience turns out to be a very important factor” (P5, FG1)
“We don't go out of our way with this, we get used to it” (P4, FG1)

It is also important to point out that some of the participants considered that all healthcare professionals should have access to supervision services as the emotional impact of same healthcare areas, like paediatrics or oncology, is also very significative:

“It is not a specific question of genetics, there are colleagues here who are closer to pediatric oncology, pediatric palliative care ... more sensitive areas and in which medical genetics is also included ... giving bad news, knowing how to deal with bad news (…).” (P5, FG2)

4. Discussion

Within the specificities of genetic counselling, considering its inherent emotional impact and the challenges, this study conducted in Portugal set out to achieve pivotal objectives. Firstly, it aimed to ascertain whether and how genetic counselling practice is supervised in Portugal. Secondly, it identified the daily routines, challenges, and impacts of genetic counselling processes that warrant exploration within a supervision framework. Lastly, it pinpointed the critical factors necessary for the successful implementation of effective supervision within the genetic counselling profession. By trying to accomplish these objectives, this study sheds light on the practice of genetic counselling in Portugal and its relationship with supervision, contributing to a deeper and more effective understanding of this crucial field.

None of the participants in this study have had or currently have access to genetic counselling supervision as a routine, however, almost all declared that they, very frequently, they felt a need to explore their feelings and to verbalize some of the difficulties they face in genetic counselling consultations.

In fact, although GCS is mandatory in some European countries, we still find in the European continent a huge heterogeneity with some of the professionals disregarding this practice, not even knowing it is benefits and how it can help them to continually improve their skills as a professional (Pestoff et al., 2018b). A European study, published in 2023 (Paneque et al., 2023), concluded that only 34% of the respondents (in a universe of 100 respondents from 18 European countries) had access to genetic counselling supervision. In the same study, researchers reached the conclusion that country of origin, the existence of a regulation

system and years of experience were factors that influenced not only the access but also the characteristics of the GCS (Paneque et al., 2023).

Once again, we can easily understand this fact if we attend the huge heterogeneity regarding genetic counselling development worldwide (Paneque et al., 2023). Across Europe we still can't find a clear and homogeneous view of what roles and skills these professionals should possess (Cordier et al., 2012; Pestoff et al., 2018a; Skirton et al., 2015), and is common to find a significant heterogeneity of tasks, with different degrees autonomy in distinct parts of the globe (Pestoff et al., 2018b; Ormond et al., 2019; Paneque et al., 2017).

4.1. Genetic counselling supervision, clinical supervision, and informal support: doubts, confusions, and assumptions

Genetic counselling supervision is still not a well-recognized area among professionals who practice genetic counselling in Portugal, resulting in many doubts about the concept and practical aspects of genetic counselling supervision. In fact, this is a relatively new field all over the world, although, in some countries, it is considered mandatory for the practice of genetic counselling (Middleton et al., 2007).

The lack of familiarity with the concept of genetic counselling supervision may explain the confusion surrounding its definition. It is worth noting that this study revealed a significant misconception between the concepts of clinical supervision and genetic counselling supervision. Clinical supervision primarily centres on the clinical aspects of processes and is based on a supervisory relationship aimed at enhancing the technical and theoretical skills of junior individuals, with the supervisor serving as a gatekeeper (Bernard and Goodyear, 2008). Clinical supervision plays a crucial role in various medical and mental health courses (Higgins et al., 2013), which naturally leads many healthcare professionals to associate genetic counselling supervision with clinical supervision.

In this study, when participants were asked whether they had access to Genetic Counselling Supervision (GCS), many of them reported that they did not personally receive GCS but instead provided supervision to less experienced professionals. They helped these junior professionals address difficulties improving their practice, which closely resembles the principles of clinical supervision.

Strategies regarding informal verbalization with peers, like having lunch or taking a break during practice, case discussion in service meetings and recurring to the service psychologist were some of the practices that participants wrongly associated to GCS, overlooking its unique characteristics, such as regularity, trained supervisors with specific skills, and the intentional nature of the practice.

This fact suggests that it may be important to provide clear explanations and guidance to professionals regarding these domains, highlighting the characteristics and specificities of genetic counselling supervision.

4.2. We did not know that existed, but we realize we needed it!

When the concept of genetic counselling supervision was clearly presented, the participants expressed their prior lack of knowledge about the subject, but also manifested a clear necessity for a service of this kind. In fact, many even underlined that they had already had previous discussions within their services regarding the need for such support, highlighting numerous potential benefits. This recognition of potential benefits may serve as a positive indicator for the implementation of genetic counselling supervision in Portugal. Many professionals stated their willingness to utilize such a service if it were available, as they experience a daily need to explore and express the challenges, they encounter in genetic counselling. In a previous study by Paneque et al. (2023), professionals already had highlighted the need for access to GCS that could assist them in managing the stressful and emotionally challenging aspects of genetic counselling (Paneque et al., 2023).

The need for emotional support among professionals became even more evident as participants shared the coping mechanisms they employ to manage the emotional burden, demanding situations, and limitations they face in their daily practice. Strategies such as substance abuse, alcohol consumption, isolation, and disengagement from the counselling processes were mentioned, underscoring the significant emotional impact of genetic counselling.

Thus, although the literature on this subject is limited, existing empirical data strongly support the need for genetic counselling supervision. Previous studies have identified some key areas that warrant attention, such as ethical dilemmas, family dynamics, and the thoughts and feelings of genetic counselling professionals during and after GC (Higgins et al., 2013). Additionally, the studies carried out point the need to empirically define skills as a way of improving GC effectiveness (Higgins et al., 2013).

The emotional impact of GC coupled with the limited guidelines on processes and procedures, underscores the urgent need for GCS, and makes even more urgent to ensure uniform standards for safe and effective practice (McAllister et al., 2016).

Additionally, participants focused on professional fatigue leading them to do the consultations always in the same way, regardless of the patient characteristics, or to avoid entering in dialogue or further explanations with patients, even when they feel it would be important. As this is an emotionally impactful job that is also very repetitive, it is essential to find tools that allow professionals to reflect on themselves and their practices, working in a logic of continuous improvement and preventing burnout. One of the key advantages of GCS is precisely related with the prevention of burnout (<https://www.ebmj.eu>). Since supervision is a practice that emphasizes the emotional and psychosocial aspects of genetic counselling professionals (Evans, 2006; Hawkins and Sholet, 2000), providing support and a platform for them to contemplate their own emotions and responses throughout the various processes they engage in (McCarthy et al., 2003), it becomes evident why Genetic Counselling Supervision (GCS) can play a vital role in preventing burnout.

4.3. Genetic counselling supervision: one size does not fit all

Our study pointed out that when asked about how could GCS be structured - individual or in a group, periodicity, duration, characteristics of the supervisor - the participants in this study had no clear opinion about it. This can be explained by the unfamiliarity with the concept.

However, a convergent group of participants considered that GCS should be operationalized according to the professional characteristics, considering that it should not exist a "one size fit all", but that supervision should also be tailored to the characteristics and specificities of the professionals and the services they work in. In fact, one of the participants even stated that "if we are all different, so should GCS be", stating that the type of supervision should adapt to the characteristics of the professional.

Indeed, several studies have found that combining individual genetic counselling supervision (GCS) with group GCS yields the best results. Group sessions are commonly utilized to address internal departmental issues such as case distribution and allocation of hours, while individual sessions allow for a more in-depth exploration of the personal aspects of professionals, including their thoughts, feelings, and the pressures they experience during genetic counselling. By combining both types of supervision, a broader and more comprehensive spectrum of support and guidance can be achieved (Middleton et al., 2007).

The supervisor characteristics were another topic where we found no consensus. If some of the participants considered that it would be mandatory for the supervisor to have a genetic background, others focused more on formal supervision skills, while others considered that the best profile would be a match of both these characteristics.

In fact, the EBMG recommendation for supervisors points out that

they should be professionals with experience and training in supervisory skills and that genetics knowledge is not mandatory, once the focus in GCS is related to the counselling itself, and not with the information or theme that is addressed.

It was also interesting to attend to the fact that all the participants considered that an external professional should carry out supervision. This comes in line with AGNC (2007) recommendations: external supervision blurs issues related to hierarchies and increases the likelihood of growth processes (AGNC, 2007). Also, external supervision seems to contribute to a greater disclosure of the professional, once he feels safe to explore different dimensions of his practice (Scanlon et al., 1997).

Even though some participants believed that the emotional impact of genetic counselling is comparable to that of other healthcare professions and should not be treated differently, there was a consensus among both focus groups that genetic counselling professionals should have access to GCS if they feel the need for it. Existing literature supports this notion, highlighting the importance of supervision for professionals in ensuring effective and high-quality genetic counselling practice, regardless of the specific structure or model employed (Middleton et al., 2007).

4.4. Genetic counselling supervision: is this luxury a necessity?

At some point, the discussion in the focus group was not about the relevance of GCS but about the urgent changes that need to be introduced in medical genetics in Portugal and the obsolete structure of genetic services. Given the actual panorama - large waiting lists, tired professionals experiencing burnout and working overtime, limited investment in research, and an outdated referral network - some participants considered that having a structured supervision service would be a "luxury".

Given these structural limitations and the need for improvements in fluidity and effectiveness in genetic counselling in Portugal, it is understandable why GCS is seen as both a necessity and a luxury. It is a necessity due to the inherent characteristics of GC and its impact on professionals. However, it is also seen as a luxury given the current state of genetic services in Portugal and the urgent changes that professionals deem essential for enhancing service quality. Despite these challenges and the pressing needs for structural improvements in genetic services, professionals considered that implementing GCS would be an excellent starting point, as it would be an area where investment and progress could be made in the short term (De Castro et al., 2016; Petersen et al., 2019; Thomas et al., 2020).

5. Conclusions

This paper highlights the unfamiliarity of health professionals with the genetic counselling supervision concept while, at the same time, underlines the need the professionals feel for a support of this kind, given the huge emotional impact of GC, the need to prevent burnout and the relevance of a continuous professional development and improvement of GC practice.

While underlining the growing need for awareness of genetic counselling supervision and the relevance it has for a safe and effective genetic counselling practice, this research also points out the need for more research in this area and the urge for uniform guidelines that can be used across Europe. The expansion of genetic testing and the mainstream innovation in this area make it even more important to have professionals who are not only well trained but that also have psychosocial support so that they can rise to the challenges they face daily.

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Ethical approval

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CRediT authorship contribution statement

Lídia Guimarães: Study design, data collection and interpretation, writing of the manuscript. **Ruxanda Baião:** Critical review of the work, focus group note-taker, english translation, approval of the final version of the manuscript. **Catarina Costa:** Critical review of the work, focus group note-taker, approval of the final version of the manuscript. **Marina Lemos:** Study design, Critical review of the work, approval of the final version of the manuscript. **Margarida Rangel Henriques:** Critical review of the work, approval of the final version of the manuscript. **Milena Paneque:** Study design, data collection and interpretation, critical review of the work, approval of the final version of the manuscript.

Declaration of competing interest

All authors declare that they have no conflicts of interest.

Data availability

The data that has been used is confidential.

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