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Portuguese Public Attitudes Regarding Contact Between Healthcare Professionals and Patient's Relatives for Genetic Risk Disclosure: A Cross-Sectional Study

Atitudes do Público Português Relativas ao Contacto Direto entre Profissionais de Saúde e Familiares do Doente para Divulgar Risco Genético: Um Estudo Transversal

Iara RIBEIRO✉[1], João TAVARES[2], Liliana SOUSA[3], Álvaro MENDES[4,5]
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ABSTRACT

Hereditary diseases can affect patients and their biological relatives, who may be at risk of developing these conditions or transmitting them to their descendants. This study explores the attitudes of a sample of Portuguese people towards receiving information about genetic risks and policies on the disclosure of such risks. An online survey using 5-point Likert scale statements (from 1 – "totally disagree" to 5 – "totally agree") was distributed through social media and in public places, resulting in a sample of 1034 respondents with a mean (M) age of 38.58 (standard-deviation, SD = 14.91) years old; 75.4% were women, 74.4% had higher education, and 55.5% were childless. The main findings suggest that participants strongly prefer to be informed about genetic risks by a doctor rather than not being informed at all (M = 4.75; SD = 0.67), are less supportive of learning of genetic risks first by a close relative (M = 3.94; SD = 1.11), favor legislation allowing healthcare professionals to contact them even if their relatives do not wish to disclose the information (M = 4.47; SD = 0.87), and are less favorable towards laws requiring individuals to inform their direct family members of their genetic risks (M = 3.88; SD = 1.27). These findings could inform ongoing discussions about the roles and responsibilities of healthcare professionals in conveying relevant information to at-risk relatives.

Keywords: Communication; Confidentiality; Family; Genetic Counseling; Genetic Predisposition to Disease; Health Personnel/ethics; Professional-Patient Relations/ethics

RESUMO

As doenças hereditárias podem afetar os pacientes e seus familiares biológicos, que podem estar em risco de as desenvolver ou transmitir aos seus descendentes. Este estudo explora as atitudes de uma amostra da população portuguesa em relação à receção de informação sobre riscos genéticos e às políticas de divulgação desses riscos. Um questionário *online*, utilizando afirmações numa escala de Likert de 5 pontos (de 1 – "discordo totalmente" a 5 – "concordo totalmente") foi divulgado nas redes sociais e em locais públicos, resultando numa amostra de 1034 inquiridos, com uma média (M) de idades de 38,58 (desvio-padrão, DP = 14,91) anos, 75,4% mulheres, 74,4% com o ensino superior e 55,5% sem filhos. Os principais resultados sugerem que os participantes prefeririam ser informados sobre os riscos genéticos por um médico a não serem informados (M = 4,75; DP = 0,67), são menos favoráveis a serem informados sobre o seu risco genético primeiro por um familiar próximo (M = 3,94; DP = 1,11), apoiariam uma lei que permitisse serem informados através do contacto direto de profissionais de saúde, mesmo que os familiares não quisessem partilhar a informação (M = 4,47; DP = 0,87); e são menos favoráveis a uma lei que obrigue os pacientes a informar os seus familiares diretos sobre o seu risco genético (M = 3,88; DP = 1,27). Estes resultados poderão contribuir para as discussões em curso sobre os papéis e responsabilidades dos profissionais de saúde na transmissão de informação relevante aos familiares em risco.

Palavras-chave: Aconselhamento Genético; Comunicação; Confidencialidade; Família; Pessoal de Saúde; Predisposição Genética para Doença; Relações Profissional-Doente

Hereditary diseases can significantly affect both patients and their genetic relatives.[1] In Portugal, as in most countries, current practice encourages probands to inform relatives about increased genetic risks, particularly for conditions with effective surveillance, prevention and treatment. However, relatives may not always receive this information,[2] preventing them from taking preventive actions or mitigating future morbidity.

Portuguese legislation stipulates that physicians must inform patients about risks to their relatives while ensuring genetic information is handled in compliance with medical confidentiality.[3] However, no specific guidelines or laws regulate the disclosure of genetic information to patients' relatives. In contrast, some countries (e.g., the Netherlands,[4] France[5]) have implemented legal frameworks allowing healthcare professionals (HPs) to directly notify at-risk relatives, provided the patient consents. International studies on public attitudes toward genetic risk disclosure reveal mixed findings, ranging from strong support for direct contact by HPs[6,7] to a preference for family-mediated disclosure.[8] This study explores the attitudes of a sample of Portuguese people toward receiving genetic risk information and policies governing its disclosure.

This observational, cross-sectional study is part of a larger project approved by the i3S Committee for Ethical and Responsible Conduct of Research (Ref. N18/2023).

1. Department of Education and Psychology. Universidade de Aveiro. Aveiro. Portugal.
2. CINTESIS@RISE. School of Health Sciences. Universidade de Aveiro. Aveiro. Portugal.
3. CINTESIS@RISE. Department of Education and Psychology. Universidade de Aveiro. Aveiro. Portugal.
4. Centre for Predictive and Preventive Genetics (CGPP). Institute for Molecular and Cell Biology (IBMC). Universidade do Porto. Porto. Portugal.
5. Instituto de Investigação e Inovação em Saúde (i3S). Universidade do Porto. Porto. Portugal.
✉ **Autor correspondente:** Iara Ribeiro. iararibeiro1@ua.pt
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Context and sample

A non-random convenience sample was used. The inclusion criteria were adults residing in Portugal. From a total of 1408 respondents, 374 were excluded (19 non-residents and 355 with incomplete questionnaire). The final sample comprised 1034 participants (response rate: 78.68%) (Table 1).

Instrument

Sociodemographic variables included gender, age, level

Table 1 – Sociodemographic characteristics of the sample (n = 1034)

Variables	n (%)
Gender	
Man	244 (23.6)
Woman	780 (75.4)
Non-binary	9 (0.9)
Other	1 (0.1)
Marital status	
Single	496 (48.0)
Married/cohabiting	446 (43.1)
Divorced/separated	79 (7.6)
Widowed	13 (1.3)
Area of residence	
Urban	840 (81.2)
Rural	194 (18.8)
Residential territorial unit	
North	479 (46.3)
Centre	236 (22.8)
Lisbon Metropolitan Area	226 (21.9)
Alentejo	35 (3.4)
Algarve	15 (1.5)
Azores Autonomous Region	23 (2.2)
Madeira Autonomous Region	20 (1.9)
Formal education	
None	1 (0.1)
5 to 6 years	19 (1.8)
7 to 9 years	17 (1.6)
Secondary (10 to 12 years)	228 (22.1)
Higher	769 (74.4)
Children	
Yes	460 (44.5)
No	574 (55.5)
Medical appointment for a hereditary disease	
Yes	210 (20.3)
No	709 (68.6)
I do not know	115 (11.1)
	M (SD; Min - Max)
Age (years)	38.58 (14.91; 18 - 81)
Number of children	0.78 (0.995; 0 - 5)

n: sample size; %: percentage; M: mean; SD: standard deviation; Min: observed minimum; Max: observed maximum.

of education, marital status, number of children, area of residence, and history of medical appointments related to a hereditary disease. A questionnaire was adapted from a previous study,[8] consisting of eight statements where participants indicated their level of agreement on a five-point Likert scale (from 1 = "completely disagree" to 5 = "completely agree"). It included questions on i) genetic testing, the receipt of information about genetic risks and the sharing of such information with family members, and ii) practice and policy approaches related to genetic risk disclosure. Here we focus on the latter.

Data collection

Data collection occurred from December 2023 to March 2024. The study was advertised through social media (via link, poster, and QR code) and in public places in Aveiro, Porto and Lisbon (flyers with QR code). The questionnaire was self-completed using the LimeSurvey platform. Access to the questionnaire was granted only after participants provided informed consent, following a review of study information, including research team contacts and a link to access the study results.

Data analysis

Frequencies and percentages describe categorical variables (gender, area of residence, education, marital status, number of children, and medical appointments for hereditary diseases). Means (M) and standard deviations (SD) describe age and number of children (discrete variables). Since normality was not confirmed by the Kolmogorov-Smirnov test, non-parametric tests (Mann-Whitney and Kruskal-Wallis) were used. Correlations between variables were tested using Spearman's correlation coefficient. Data were analyzed using SPSS statistical software 28.0 (SPSS Inc., USA).

Participants

The 1034 participants had a mean age of 38.58 (SD = 14.91) years (range: 18 - 81 years); 75.4% were women, 48% were single, and 43.1% were married or cohabiting. The majority resided in urban areas (81.2%), had completed higher education (74.4%), and were childless (55.5%). Respondents lived mainly in the North (46.3%), Centre (22.8%) or in the Lisbon Metropolitan Area (21.9%) (Table 1).

Receiving information about genetic risk

The highest mean preference was for being informed by a doctor rather than not being informed (M = 4.75; SD = 0.67), followed by a preference for being informed by a distant relative rather than not being told (M = 4.31; SD = 1.06), and for being informed first by a doctor rather than by a distant relative (M = 4.21; SD = 1.06). The lowest mean preference was for being informed first by a close relative (M = 3.94; SD = 1.11) (Table 2). No significant differences or associations were observed across sociodemographic variables.

Policies on genetic risk disclosure

The highest mean preference was for supporting a law allowing HPs to contact patients' relatives directly, even if relatives choose not to inform them (M = 4.47; SD = 0.87). This was followed by support for authorizing HPs to access personal data from population registers (M = 4.40; SD = 0.90) and contact details (M = 4.28; SD = 1.01) to inform individuals of genetic risks (Table 2). The lowest mean preference was for supporting a law obligating patients to inform their direct relatives of their risk (M = 3.88; SD = 1.27).

Significant differences were observed based on gender, marital status, formal education, and having had a medical appointment for a hereditary disease. Specifically, the preference for authorizing HPs to obtain personal data from population registers was significantly higher among women than men (U = 87,649.0; $p = 0.032$), divorced participants compared to singles [Mann-Whitney test with Bonferroni correction, H(2) = 6.899; $p = 0.032$; $\varepsilon^2 = 0.005$], participants with less than nine years of formal education compared to those with secondary and higher education [Mann-Whitney test with Bonferroni correction, H(2) = 9.450; $p = 0.009$; $\varepsilon^2 = 0.010$], and those with children compared to those without (U = 120,181.5; $p = 0.004$).

Women also showed significantly higher support than men for authorizing HPs to access personal contact details to inform them of genetic risks (U = 85,446.0; p = 0.007). Support for a law obligating patients to inform direct relatives was significantly higher among divorced participants compared to singles [Mann-Whitney test with Bonferroni correction, H(2) = 6.120; $p = 0.047$; $\varepsilon^2 = 0.040$]. Participants who had not had a medical appointment for a hereditary disease showed significantly higher preferences than those who answered "I do not know" [Mann-Whitney test with Bonferroni correction, H(2) = 10.045; $p = 0.007$; $\varepsilon^2 = 0.007$] (Table 2).

This study is the first to explore public attitudes toward genetic risk disclosure in Portugal. The main findings suggest that the sample of Portuguese people prefers being informed of genetic risks directly by a HP rather than by a family member, support policies that facilitate dissemination of genetic risk information by HPs, and is less supportive of policies that legally require patients to inform relatives of increased genetic risk. A previous study with people living with hereditary diseases in Portugal similarly reported widespread acceptance of HP-mediated direct approaches.[9] These findings are consistent with

Table 2 – Receiving information about genetic risk and policies on genetic risk disclosure, and sociodemographic variables (significant differences only)

	M (SD)	Statistical result	M (SD)	Statistical result	M (SD)	Statistical result	M (SD)	Statistical result
I would prefer:								
	To be informed by a doctor rather than not be informed.		To be informed by a distant relative rather than not be told.		The first communication to be made by a close relative.		The first communication to be made by a doctor rather than a distant relative.	
	4.75 (0.67)		**4.31 (1.06)**		**3.94 (1.11)**		**4.21 (1.06)**	
I would support/authorize:								
	Healthcare professionals to access my data from population registers to inform me.		Sharing my personal contact details to enable me to be informed		A law that would allow health professionals to contact me, even if my relatives didn't wish to inform me.		A law that would require patients to inform their direct family members.	
	4.40 (0.90)		**4.28 (1.01)**		**4.47 (0.87)**		**3.88 (1.27)**	
Gender[a]								
Men (244)	4.32 (0.91)	U = 87649 p = 0.032	4.14 (1.08)	U = 85446.0 p = 0.007				
Women (780)	4.42 (0.90)		4.33 (0.98)					
Marital status[b]								
Single (496)	4.40 (0.83)	$H(2)$ = 6.899 p = 0.032					3.87 (1.23)	$H(2)$ = 6.12 p = 0.047
Married (446)	4.37 (0.97)						3.82 (1.32)	
Divorced (79)	4.56 (0.93)						4.20 (1.23)	
Formal education[c]								
Up to 9 (36)	4.83 (0.46)	$H(2)$ = 9.45 p = 0.009						
Secondary (228)	4.64 (0.85)							
Superior (769)	4.38 (0.91)							
Children								
Yes (460)	4.45 (0.92)	U = 120181.5 p = 0.004						
No (574)	4.36 (0.88)							
Have had a medical appointment about a hereditary disease								
Yes (210)							3.90 (1.23)	$H(2)$ = 10.045 p = 0.007
No (709)							3.91 (1.30)	
I do not know (115)							3.72 (1.28)	

[a]: non-binary gender not considered, n = 9;
[b]: Widowers not considered, n = 13;
[c]: No schooling not considered, n = 1;
M: mean; SD: standard-deviation; U: Mann-Whitney test; H: Kruskal-Wallis test.
Spearman's correlation between all the statements and age and number of children were all weak (r_s < 0.1), and statistically significant (p < 0.05). In all the statements, the minimum and maximum observed values were 1 and 5.

international public opinion.[6-8] The limitations of this study include the use of a convenience sample, leading to an overrepresentation of women, individuals with higher education, and those living in urban areas. The recruitment method also introduced a bias towards individuals more engaged with online platforms and social media. Future research should include additional questions and aim to collect a representative sample. Our findings suggest that the Portuguese public favors a more proactive approach from the healthcare system in notifying patient's relatives of genetic risks, compared to current practice. Broader discussions are needed on how to appropriately cascade genetic risk information, involving all relevant stakeholders.[1,9,10]

PREVIOUS AWARDS AND PRESENTATIONS

Ribeiro I, Tavares J, Freixo JP, Sousa L, Mendes A. Disclosure of genetic risk in the family: Attitudes of the general Portuguese population. Hybrid poster presentation from the 56th European Society of Human Genetics (ESHG) Conference; 2024 Jun 1 - 4; Berlin, Germany.

Ribeiro I, Tavares J, Sousa L, Mendes A. Portuguese public attitudes towards genetic risk disclosure support

direct contact by healthcare professionals with family members. Poster presentation from the 28th Portuguese Society of Human Genetics (SPGH) Meeting; 2024 Dec 5 - 7; Porto, Portugal.

AUTHOR CONTRIBUTIONS

IR: Data collection, writing of the manuscript.
JT: Data analysis.
LS, AM: Study design, critical review of the manuscript.
All authors approved the final version to be published.

PROTECTION OF HUMANS AND ANIMALS

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the Helsinki Declaration of the World Medical Association updated in October 2024.

DATA CONFIDENTIALITY

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

COMPETING INTERESTS

AM acknowledges support from FCT through the research contract CEECIND/02615/2017.

All the other authors declare no competing interests.

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