



Psychosocial needs and preferences of individuals with hereditary cancer syndromes: Practice implications for the healthcare responsiveness

Eliana Silva, Pedro Gomes, Paula M. Matos, Eunice R. Silva, João Silva, & Célia M. D. Sales





Introduction



Hereditary cancer syndrome

- About 5% to 10% of cancers are strongly related to an **inherited genetic mutation**.
- These genetic mutations are associated with more than **50 hereditary cancer syndromes** (National Cancer Institute, 2017)
 - Hereditary breast and ovarian cancer syndrome (HBOC)
 - Hereditary non-polyposis colorectal cancer (HNPCC) - Lynch Syndrome (LS)
 - Familial adenomatous polyposis (FAP)
- **Early identification and prophylactic treatment** “potentially reduce the incidence/prevalence and lessen morbidity and mortality” (Gandy and Rodning, 2011, p. 46)

Cancer genetic risk assessment and genetic counseling





Cancer risk assessment and counseling

- **Personalized management plan** (Blix, 2014)
- Individuals with hereditary cancer syndromes usually have a long-term risk management process, which requires a **responsive healthcare system** (Bleich et al., 2009)
- Most researches used patient-reported outcome measures through **quantitative studies** (e.g., D'Andrea et al., 2018; Nilsson et al., 2019)
 - **qualitative studies** (Etchegary et al., 2015; Samuel et al., 2017)
 - specific hereditary cancer syndrome
 - specific domain of healthcare responsiveness
 - specific medical procedures





The present study

- This is the first qualitative study that focuses on the care experiences of pathogenic/likely pathogenic variant carriers along with **cancer risk assessment, genetic counseling, and follow-up surveillance.**
- Two specific aims:
 - to describe the **participants' experiences of care**
 - to **identify their psychosocial needs and preferences**



Method



Participants

Characteristics	<i>N</i> = 13
Mean age (range)	35.08 (22-53)
Gender	
Male	7
Female	6
Hereditary cancer syndromes	
HBOC	3
HDGCS	4
LS	5
FAP	1
Risk management approaches	
Prophylactic surgery	9
Enhanced screening	4



Procedure

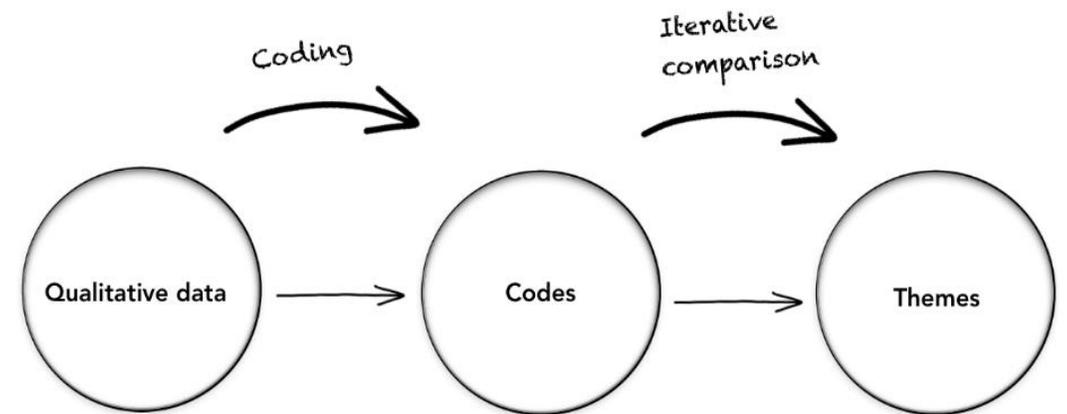
- Participants were attending **risk management consultations** at a Portuguese Oncology Hospital.
- This study was part of the **TOGETHER research project** (POCI-01-0145-FEDER-030980).
 - Approved by the **Hospital's ethics board** (informed consent was obtained from participants)
- Data were collected through a ninety-minute **semi-structured retrospective interview**.
- We adopted a **participatory approach**
 - **staff panel** (2 clinical oncologists)
 - **user panel** (n = 6; 83% female)





Data analysis

The interviews were transcribed in full and analyzed using **thematic analysis** (Braun and Clarke, 2006)





Results



Main Themes



Quality of clinical
information



Clinical relationship



Psychological
support



Healthcare
organization
processes





Quality of clinical Information

1) Need of information

2) Lack of understanding of
the information

Unmet information needs: about genetic testing and the postoperative adaptation

“We have to decide to do or not to do the genetic testing, but without knowing exactly what that means” [15, HDGC]

Evidence- and experience-based information: the disease, side effects, and adaptation

“(How) do you feel without a stomach? Do you feel an emptiness inside?” [...]
These are sometimes little stupid questions that we have, but even physicians can’t answer because they have the stomach. [17, HDGC]



Quality of clinical Information

1) Need of information

2) Lack of understanding of
the information

Some participants looked for additional information on the **Internet** and others contacted **healthcare professionals**

“I didn’t go on the internet, because on the internet there is nothing reliable related to this.” [I1, HBOC]



Clinical Relationship

1) Emotional bond and long-term relationship

2) Shared decision-making

3) Person-centered care

Being seen by the same physician to develop a **relationship of trust**

“If I have to decide [...] the best person [...] to give me some support or try to help me in any decision was my oncologist because she has been with me all these years and she is a person I have confidence in [...]” [I12, LS]



Clinical Relationship

1) Emotional bond and long-term relationship

2) Shared decision-making

3) Person-centered care

Participants appreciate being **actively involved in decisions** about treatment, **being informed** about their condition, and having **the freedom to decide** about surgery

“I felt that I was not being pressured [...]. She [the psychologist] basically letting the decision up to me” [15, HDGC]



Clinical Relationship

- 1) Emotional bond and long-term relationship
- 2) Shared decision-making
- 3) Person-centered care**

Responsiveness to participants' needs: availability of prescribing medication

Interest and communication beyond the syndrome

“Talking about issues not directly related to the clinical information helps a lot [conversation between participant and physician about attending a concert after receiving bad news]. I know it could be a strategy of the physicians to ease the situation, but it helped a lot” [I7, HDGC]

Healthcare professionals use the **label “patient”** to communicate with mutation carriers

“I’m not “sick”, I just don’t have a stomach” [I7, HDGC]

Psychological Support



1) Individual psychological support

2) Psychological support for relatives

3) Suggestions of new approaches for support

Before undergoing genetic testing: to increase understanding and aid decision-making

After receiving results: to help cope with emotional reactions and psychological impact

Physicians should **refer to psychological support**

“Because often the person is not even aware that they are in need [...] So I think it should be a standard procedure.” [12, HBOC]



Psychological Support

1) Individual psychological support

2) Psychological support for relatives

3) Suggestions of new approaches for support

On the **psychological distress of prophylactic surgery** (its implications for the couple)

Finding strategies (how and when) to communicate with their children the future need for genetic testing

Psychological Support

- 
- 1) Individual psychological support
 - 2) Psychological support for relatives
 - 3) Suggestions of new approaches for support

More frequent psychological support

Group meetings with the inclusion of the family

Support from a **multidisciplinary team** (i.e., mental health professionals, physicians, nurses)

Telephone counseling

Online support using remote videoconferencing



Healthcare organization processes

- 1) Inclusion of individuals' family
- 2) Time-frames of medical procedures
- 3) Ombudsman service
- 4) Social benefits
- 5) Information resources
- 6) Surveillance frequency

To facilitate **the presence of family members** in the care setting to support hospitalization

Healthcare organization processes

1) Inclusion of individuals'
family

**2) Time-frames of medical
procedures**

3) Ombudsman service

4) Social benefits

5) Information resources

6) Surveillance frequency

Be informed about the results of medical exams as soon as possible

Healthcare organization processes

- 
- 1) Inclusion of individuals' family
 - 2) Time-frames of medical procedures
 - 3) Ombudsman service**
 - 4) Social benefits
 - 5) Information resources
 - 6) Surveillance frequency

To provide the opportunity to have one person in the hospital **who serves the rights and interests** of individuals

Healthcare organization processes

- 1) Inclusion of individuals' family
- 2) Time-frames of medical procedures
- 3) Ombudsman service
- 4) Social benefits**
- 5) Information resources
- 6) Surveillance frequency

Financial support

"If I hadn't had lymphoma, I would have had to pay for all the medical tests.

I didn't have that support" [I11, LS]

Justifications for absences from work/university

"I really felt the need to explain my health condition [to my teacher] so that she could realize that I was not missing classes because I wanted to" [I5,

HDGC]

Healthcare organization processes

- 
- 1) Inclusion of individuals' family
 - 2) Time-frames of medical procedures
 - 3) Ombudsman service
 - 4) Social benefits
 - 5) Information resources**
 - 6) Surveillance frequency

To receive information about **available customer services** (e.g., counseling support)

Healthcare organization processes

- 
- 1) Inclusion of individuals' family
 - 2) Time-frames of medical procedures
 - 3) Ombudsman service
 - 4) Social benefits
 - 5) Information resources
 - 6) Surveillance frequency**

To have **more frequent medical appointments and time** from physicians

“Since the postoperative period, especially [...] Because unfortunately, people [physicians] have 10 minutes to be with us, and the 10 minutes start counting since they enter the service, they open our clinical record [...] There is no time to ask a question” [I1, HBOC]



Discussion

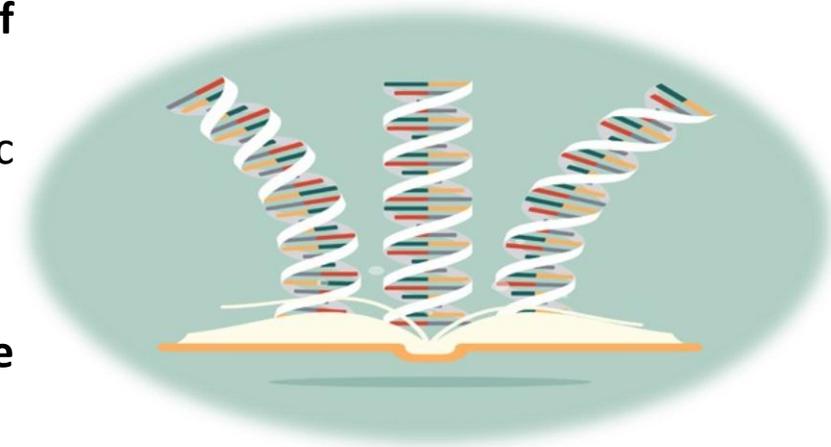


- The psychosocial needs and preferences include the **families** of mutation carriers' and are not limited to their **clinical condition**, but also to their **personal identity**.
- Individuals have **different needs along with all the specific phases** of adaptation.



Practice Implications

- 1) Provision of **more informational support** and **availability of support resources**
- 2) Integration of **multi-family discussion groups** into genetic counseling
- 3) The **availability** and **clinical referral of psychological support**
- 4) The provision of **long-term, individual- and family-centered care** that meets the health and personal needs of individuals





Limitations

- The participants' experiences **may represent little diversity**: they are mainly associated with one healthcare center.
- **The retrospective nature of the data collected**: some participants had already undergone genetic testing or surgery a long time ago.



Conclusions

- Individuals **appreciated being informed** about their clinical condition and available preventive approaches, and **engaged in humanized, accessible, and responsive healthcare**.
- This first-hand knowledge of mutation carriers' experiences is particularly valuable for **healthcare professionals, and settings become more responsive** to these individuals who need clinical surveillance throughout their lives.



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