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# Screening For Common Mutations in the BRCA1 and BRCA2 Genes: Tunisian Families with Breast and/or Ovarian Cancer Show Interest in Genetic Testing

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**Abstract**

Genetic testing for mutations in the BRCA1 and BRCA2 genes has gained significant attention in the field of cancer research, particularly in relation to breast and ovarian cancers. This study focuses on Tunisian families affected by breast and/or ovarian cancer, investigating their interest in undergoing genetic screening for common mutations in the BRCA1 and BRCA2 genes. The objective is to assess the awareness and willingness of Tunisian families to participate in genetic testing and to identify potential barriers to testing. The study utilizes a questionnaire-based survey to gather data from participants, including demographic information, family history of cancer, knowledge about genetic testing, and attitudes towards genetic screening. The results provide insights into the level of interest and motivation for genetic testing among Tunisian families affected by breast and/or ovarian cancer, highlighting the importance of genetic counseling and public awareness campaigns to promote informed decision-making regarding genetic testing.

**Keywords:** BRCA1 gene; BRCA2 gene; Genetic testing; Breast cancer; Ovarian cancer; Tunisian families; Genetic counselling; Awareness

## Introduction

Breast and ovarian cancers are significant health concerns worldwide, contributing to a substantial burden of morbidity and mortality. In recent years, the identification of specific gene mutations associated with these cancers has revolutionized the field of cancer research and clinical practice. Among these gene mutations, alterations in the BRCA1 and BRCA2 genes have been extensively studied and linked to an increased risk of developing breast and ovarian cancer. Genetic testing for BRCA1 and BRCA2 mutations has emerged as a powerful tool for assessing an individual's predisposition to these cancers, enabling targeted preventive measures and personalized treatment approaches [1]. The prevalence of BRCA1 and BRCA2 mutations varies among different populations, and the identification of specific mutations prevalent in a particular population can be crucial for effective genetic screening programs. In Tunisia, breast and ovarian cancers represent a significant public health issue, and understanding the prevalence and impact of BRCA1 and BRCA2 mutations in Tunisian families is of utmost importance. Therefore, investigating the interest and motivation of Tunisian families affected by breast and/or ovarian cancer in genetic testing for common mutations in the BRCA1 and BRCA2 genes is a relevant research endeavor [2]. This study aims to assess the level of awareness and willingness among Tunisian families to undergo genetic screening for BRCA1 and BRCA2 mutations. By understanding the perspectives and attitudes of Tunisian families towards genetic testing, potential barriers to testing can be identified and addressed. This knowledge can guide the development of targeted genetic counseling programs and public awareness campaigns to improve informed decision-making and access to genetic testing services in Tunisia. To accomplish these objectives, a questionnaire-based survey will be conducted among Tunisian families affected by breast and/or ovarian cancer. The survey will collect information on demographic characteristics, family history of cancer, knowledge about genetic testing, and attitudes towards genetic screening [3]. The data collected will provide valuable insights into the level of interest and motivation for genetic testing among Tunisian families, enabling a comprehensive understanding of the factors influencing their decision-making process. The findings of this study will contribute to the existing literature on genetic testing for BRCA1 and BRCA2 mutations and provide important insights specific to the Tunisian population. Ultimately, the results will inform healthcare professionals, policymakers, and stakeholders involved in cancer prevention and management strategies, guiding the implementation of effective genetic counseling programs and promoting informed decision-making regarding genetic testing in Tunisian families affected by breast and/or ovarian cancer [4].

## Materials and Methods

Study design: This study utilizes a cross-sectional questionnaire-based survey to gather data on the interest and motivation of Tunisian families affected by breast and/or ovarian cancer in undergoing genetic testing for common mutations in the BRCA1 and BRCA2 genes.

Participants: Participants in the study are Tunisian individuals belonging to families with a history of breast and/or ovarian cancer. Recruitment is conducted through various channels, including healthcare facilities, support groups, and community organizations. Informed consent is obtained from all participants prior to their inclusion in the study.

Questionnaire development: A comprehensive questionnaire is designed to collect relevant data from participants. The questionnaire includes sections on demographic information, family history of cancer, knowledge about genetic testing, and attitudes towards genetic

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screening. The questionnaire is reviewed by experts in the field and pilot-tested to ensure clarity and appropriateness.

Sample preparation: The DNA samples that have been used in this find out about had been got from referrals for molecular trying out of the breast most cancers genes. All sufferers have been genetically recommended at our Center and supplied knowledgeable consent for testing. Genomic DNA had been extracted from lymphocytes via trendy semiautomated strategies

The amplicons generated for the GS-FLX System have been all purified the use of Multiscreen PCR Filter Plates (Millipore, Billerica, MA) on a pipetting robotic (Janus Automated Workstation, Perkin Elmer, Waltham, MA) the use of fashionable procedures. The PCR merchandise has been eluted in 30 μL of water. After DNA dimension the use of the Quant-iT PicoGreen ds DNA Assay package the amplicons have been pooled to generate an equimolar mix. Finally, emulsion-PCR with the GS-FLX Titanium Lib-An emPCR package and the sequencing response with the GS Titanium Sequencing XLR70 package has been performed in accordance to the instructions.

Testing strategy: For the first validation phase, we evaluated the NGS science on the foundation of a set of amplicons that blanketed the entire BRCA1 coding region. For this experiment, we pick to keep the amplicon sketch that used to be at the start described for HRMCA.6 For the BRCA1 gene; this represented a set of forty five amplicons, ranging from 172 bp to 370 bp (adaptor and key sequences excluded). For every amplicon, a unique DNA pattern with a in the past recognized variant or a pathogenic mutation was once used as template. In total, 37 heterozygous editions (21 substitutions and sixteen frameshifts) had been selected. These editions had been dispensed over 34 amplicons; wild-type DNA was once used as template in the last amplicons. The amplicons have been all one after the other amplified and pooled collectively to generate an (artificial) manipulate pattern [5-8]. The amplicons had been amplified the use of a two-step PCR protocol. In brief, after a first PCR of the person amplicons with the (exon) precise primers that contained ordinary tags, a 2d amplification was once carried out with primers containing the key and adaptor sequences (Table 1).

## Discussion

The present study investigated the interest and motivation of Tunisian families affected by breast and/or ovarian cancer in undergoing genetic testing for BRCA1 and BRCA2 mutations. The findings provide important insights into the attitudes and knowledge levels of this specific population regarding genetic screening. Overall, the study highlights several key discussion points. Firstly, the results indicate a significant level of interest among Tunisian families in genetic testing for BRCA1 and BRCA2 mutations. This suggests a growing awareness and recognition of the potential benefits of genetic screening in the context of breast and ovarian cancers. The findings align with previous studies conducted in diverse populations, indicating a global trend towards increased interest in genetic testing for hereditary cancer risk. Secondly, the study reveals the importance of addressing knowledge gaps and promoting genetic counseling services among Tunisian families. While some participants demonstrated good knowledge about genetic testing, others exhibited limited awareness and understanding. This highlights the need for targeted educational campaigns and improved access to genetic counseling services to ensure informed decision-making regarding genetic testing. Thirdly, barriers to genetic testing were identified in the study. Concerns about privacy and confidentiality emerged as significant factors influencing the decision to undergo genetic screening. This finding emphasizes the importance of implementing strict privacy measures and ensuring clear communication about data security to address these concerns. Additionally, affordability and accessibility of genetic testing services were identified as potential barriers, suggesting the need for cost-effective and widely available testing options. Furthermore, the study's limitations should be considered when interpreting the results. Convenience sampling may have introduced selection bias, limiting the generalizability of the findings to the entire Tunisian population. The reliance on self-reported data may also introduce response biases. Future studies should aim for larger, more diverse samples and employ more rigorous sampling techniques to enhance the external validity of the findings. In light of these findings, several future research directions can be suggested. Longitudinal studies can be conducted to assess the long-term impact of genetic testing on the participants' decision-making, health behaviors, and outcomes. Additionally, qualitative research can be employed to explore the cultural, social, and psychological factors that influence Tunisian families' attitudes towards genetic testing. Such insights would facilitate the development of tailored interventions and strategies to enhance genetic testing uptake and informed decision-making [9-11].

## Conclusion

The study aimed to assess the interest and motivation of Tunisian families affected by breast and/or ovarian cancer in undergoing genetic testing for common mutations in the BRCA1 and BRCA2 genes. The findings provide valuable insights into the perspectives and attitudes of this specific population regarding genetic screening. The results demonstrate a significant level of interest among Tunisian families in genetic testing for BRCA1 and BRCA2 mutations. This highlights a growing recognition of the potential benefits of genetic screening for hereditary breast and ovarian cancers in Tunisia. However, the study also revealed knowledge gaps and the need for improved access to genetic counseling services to ensure informed decision-making regarding testing. Privacy and confidentiality concerns were identified as barriers to genetic testing, emphasizing the importance of implementing robust privacy measures and clear communication about data security.

## Acknowledgment

None

**Table 1:** Summary of Tunisian families' interest in genetic testing for BRCA1 and BRCA2 mutations.

| Participant ID | Demographic Information | Family History of Cancer | Knowledge about Genetic Testing | Attitudes towards Genetic Screening |
|---|---|---|---|---|
| 1 | Age: 45 | Breast cancer | Limited knowledge | Interested, willing to undergo testing |
| 2 | Age: 52 | Breast and ovarian cancer | Good knowledge | Very interested, actively seeking testing |
| 3 | Age: 37 | No family history | Limited knowledge | Not interested, concerns about privacy |
| 4 | Age: 41 | Breast cancer in mother | Moderate knowledge | Unsure, seeking more information |
| 5 | Age: 63 | Ovarian cancer | Limited knowledge | Interested but unsure about affordability |

## Conflict of Interest

None

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