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Improving Genetic Literacy And Access To Breast Cancer Genetic Testing In Indian Healthcare Systems

Harshita Singh*
Usha Mittal Institute of Technology (UMIT), SNDT Women's University.
Santacruz, Mumbai

Harshada Jagade
Usha Mittal Institute of Technology (UMIT), SNDT Women's University.
Santacruz, Mumbai

Shraddha Rokade
Usha Mittal Institute of Technology (UMIT), SNDT Women's University.
Santacruz, Mumbai

Vishram Bapat
Databyte Systems and Software, Head of Innovari: Student Innovation and Mentorship Program, UMIT, Board of Director WISE: SNDT Incubation Center
Santacruz, Mumbai

Kanwaljeet Kaur Saluja
Usha Mittal Institute of Technology (UMIT), SNDT Women's University.
Santacruz, Mumbai

***Abstract*—**Genetic testing is a vital tool in assessing breast cancer risk, guiding treatment decisions, and improving early detection and personalized treatment. However, in India, limited genetic literacy—defined as the ability to understand and apply genetic information in healthcare decisions—combined with financial, infrastructural, and policy-related barriers, significantly restricts access to these life-saving services. Our findings show that only 10–15% of eligible patients undergo testing, and fewer than 30% of hospitals offer genetic counseling. Despite 22.5% of patients having a family history of breast cancer, awareness of BRCA mutations remains low. To improve access, we recommend integrating genetic counseling, expanding literacy programs, and implementing cost-reduction policies, ensuring early detection and personalized treatment for better patient outcomes. Such efforts will facilitate early diagnosis, improve treatment personalization, and ultimately enhance patient outcomes. Addressing these systemic issues is critical to ensuring equitable access to genetic services in India and fostering a more informed, proactive approach to breast cancer prevention and care

Keywords: Genetic Testing, Genetic Literacy, Breast Cancer, BRCA Mutations, Genetic Counseling, Healthcare Access, Personalized Medicine, India, Early Detection, Precision Oncology.

I. INTRODUCTION

Genetic testing and counseling have revolutionized the landscape of modern healthcare by providing individuals with valuable insights into their hereditary risks for various diseases [1]. These services enable early detection, proactive medical interventions, and personalized treatment strategies, significantly improving patient outcomes [2]. The growing field of medical genetics has demonstrated that many diseases, including cancer, cardiovascular disorders, neurodegenerative conditions, and rare genetic syndromes, have a strong hereditary component [3]. Identifying genetic markers through testing allows healthcare providers to tailor treatment plans and preventive measures, ultimately reducing the overall burden of chronic and life-threatening diseases .

This paper explores the importance of genetic testing in disease prevention and its potential to improve healthcare outcomes in India. It identifies the key obstacles limiting its widespread adoption, including financial, educational, and policy-related challenges. The study also examines the current state of genetic literacy among healthcare providers and patients in Indian hospitals, emphasizing the need for structured education and training programs. Moreover, the paper outlines actionable strategies to integrate genetic testing into routine healthcare, advocating for policy reforms, cost reduction measures, and public awareness campaigns. Addressing these critical issues is imperative to making genetic testing a widely accessible and affordable service,

ensuring that individuals across all socioeconomic backgrounds can benefit from advancements in medical genetics.
By implementing a multi-faceted approach that includes government intervention, industry collaboration, and large-scale awareness programs, India can pave the way for a healthcare system that prioritizes early detection, personalized medicine, and preventive healthcare. Bridging the gap between genetic advancements and practical applications will not only empower individuals to take charge of their health but will also reduce the overall burden of genetic diseases in the country's healthcare system.

II. On the importance Genetic Testing and Genetic Counseling in the Healthcare

"Affordable and accessible genetic testing is crucial in developing countries, where financial constraints often limit the use of genetic medicine" [4].

Genetic testing is defined as "the analysis of an individual's DNA to identify mutations associated with inherited conditions" [5]. When combined with genetic counseling, it provides individuals with crucial insights into their predisposition to hereditary diseases, thereby enabling them to make informed healthcare decisions. This process is particularly valuable for conditions such as breast cancer, where "mutations in BRCA1 and BRCA2 genes significantly increase an individual's lifetime risk" [6]. Similarly, hereditary cardiovascular diseases and rare genetic disorders benefit from early genetic detection, as early intervention can lead to better treatment outcomes . Genetic counseling plays a pivotal role in interpreting test results, addressing ethical concerns, and guiding patients toward appropriate medical interventions, making it an essential aspect of personalized medicine [7].

A. Benefits of Genetic Testing include*:*

- Early Detection and Prevention: According to the Centers for Disease Control and Prevention (CDC), "genetic testing allows individuals to take preventive actions before symptoms appear, such as lifestyle modifications, regular screenings, and preemptive treatments" [8]. This significantly reduces the risk of developing full-blown diseases and improves long-term prognosis [9].
- Personalized Treatment Plans: Advances in medical genetics have shown that "patients with genetic predispositions can receive tailored treatment strategies that align with their genetic makeup". Targeted therapies, such as PARP inhibitors for BRCA mutation carriers, exemplify the power of personalized medicine in optimizing health outcomes [10].
- Family Planning Guidance: Genetic testing provides valuable insights into reproductive risks, helping individuals assess the likelihood of passing hereditary conditions to future generations. Studies indicate that "genetic counseling aids prospective parents in making informed reproductive decisions, including the use of assisted reproductive technologies" [11].
- Reduced Healthcare Costs: Research suggests that "early intervention through genetic testing significantly lowers the financial burden associated with late-stage disease management" [12]. By detecting conditions at an early stage, genetic testing minimizes the need for extensive treatments, hospitalizations, and long-term medications, thereby reducing overall healthcare expenditures [13].
- Improved Psychological Preparedness: Genetic counseling plays a crucial role in alleviating anxiety by providing clear, evidence-based information about an individual's genetic risk. As Kaphingst et al. (2009) note, "genetic counseling helps individuals and families develop coping strategies and provides emotional support, preparing them mentally for potential genetic risks."[14].

However, the high cost of genetic testing in India, ranging from ₹15,000 to ₹50,000, with advanced tests such as whole genome sequencing costing even more, makes these services unaffordable for a significant portion of the population [15][16]. Financial barriers are further exacerbated by the exclusion of genetic testing from most health insurance policies, forcing patients to bear the entire cost out-of-pocket, which discourages many from opting for potentially life-saving tests [17][18]. The scarcity of government-supported genetic testing centers, particularly in rural areas, necessitates reliance on private laboratories that charge high fees, creating additional financial obstacles for lower-income groups [19][20]. Furthermore, India depends heavily on imported genetic testing kits, reagents, and specialized equipment, leading to inflated costs due to high import duties, taxes, and transportation expenses [21][22]. Unlike in developed nations, India has minimal government subsidies or financial assistance programs to make genetic testing more affordable, restricting lower-income families from accessing these crucial healthcare services [23][24]. The genetic testing industry in India remains fragmented, with few domestic manufacturers producing high-quality testing kits, while setting up a laboratory requires costly equipment, skilled personnel, and compliance with stringent regulatory standards, further driving up costs [25][26]. Additionally, government collaboration with private genetic testing firms remains limited, despite the potential for public-private partnerships to lower costs and improve accessibility [27]. Addressing these financial and infrastructural challenges through policy reforms, subsidies, and increased domestic production is crucial for ensuring equitable access to genetic testing in India.

Low public awareness remains a significant barrier to genetic testing adoption in India, as many individuals remain uninformed about its benefits and rely solely on conventional diagnostic procedures due to inadequate dissemination of information through healthcare systems [16][23]. Additionally, many physicians, particularly in rural areas, lack adequate training in genetics, leading to suboptimal recommendations for genetic testing, as continuous medical education programs on genetics remain scarce [15][25]. Cultural and social stigma further deter individuals from seeking genetic testing and counseling, as hereditary conditions are often considered taboo in some communities, and fear of societal judgment prevents individuals from acknowledging their risk factors [17][26]. Moreover, misinformation and fear contribute to the reluctance to undergo genetic testing, with concerns over discrimination in employment or health insurance coverage, as well as ethical concerns regarding genetic data privacy [21][22]. The complexity of genetic concepts, often communicated using technical jargon, further hinders public understanding, as the absence of simplified educational materials exacerbates confusion and reluctance to consider genetic testing[18][24]. Limited educational initiatives, including the minimal inclusion of genetic literacy in public health campaigns and school curricula, result in a lack of foundational knowledge about genetics among the general population, necessitating targeted awareness programs [19][20]. Furthermore, genetic literacy receives little representation in mainstream media, limiting public engagement and awareness of its importance in healthcare, as media campaigns on preventive healthcare rarely emphasize genetic risk factors [25][27]. The absence of grassroots community outreach programs further compounds these challenges, particularly in rural and underprivileged areas, where mobile health units and local advocacy programs could play a vital role in raising awareness and promoting genetic testing [15][16]. Addressing these gaps through policy-driven education initiatives, media engagement, and improved physician training is essential to enhancing genetic literacy and facilitating broader access to genetic services in India.

Additionally, genetic testing is largely confined to tertiary care centers, making it inaccessible to rural populations and limiting early detection efforts [16]. The absence of a national policy further exacerbates disparities, as regulations primarily focus on ethical concerns rather than affordability and accessibility [20]. Moreover, the lack of standardized protocols results in inconsistent testing methods and interpretations, undermining the reliability of genetic diagnoses [25]. Addressing these challenges requires expanding genetic services to primary healthcare, implementing regulatory reforms, and establishing quality assurance measures to ensure equitable and effective genetic testing in India .

B. Genetic Literacy in Indian Hospitals

Genetic literacy, defined as the awareness, comprehension, and application of genetic information by healthcare professionals and patients, remains notably low in India, significantly hindering the effective implementation of genetic testing and counseling services [22]. Studies suggest that fewer than 30% of Indian hospitals integrate genetic counseling into routine care, leaving many individuals with hereditary conditions or genetic risk factors without adequate guidance [16]. This lack of counseling often results in misinterpretation of test results, unnecessary anxiety, and missed opportunities for early intervention [17]. Despite advancements in genetic diagnostics, only 10-15% of eligible patients undergo genetic testing, primarily due to limited awareness, financial constraints, and restricted access in primary care settings [20]. The absence of widespread public health initiatives further exacerbates this issue, preventing many individuals from understanding the significance of genetic screening for hereditary conditions such as breast cancer, sickle cell anemia, and thalassemia [21]. Addressing these gaps through structured education programs, policy interventions, and improved accessibility to genetic services is essential for integrating genetic medicine into mainstream healthcare.

C. Figures and Tables

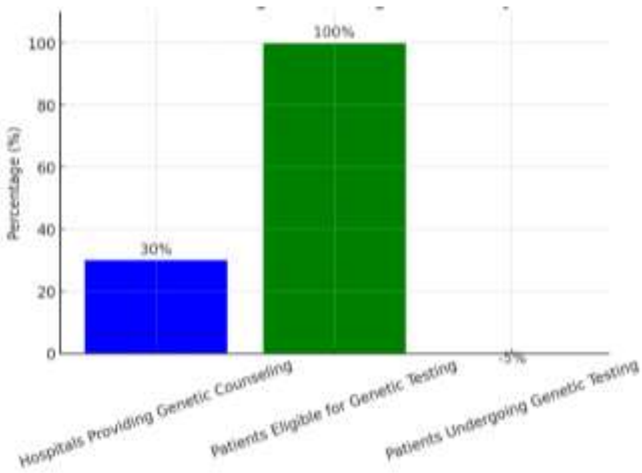

Fig 1. Genetic Counselling and Testing Accessibility in India

The survey findings highlight a significant gap in genetic literacy and access to genetic testing services in Indian hospitals. Addressing these challenges requires a multi-pronged approach, including medical education reforms, public awareness initiatives, financial assistance programs, and policy interventions.

Comprehensive Analysis of Breast Cancer Patient Data (2024)

This study collected data from 300 patients at Specialty Surgical Oncology Hospital & Research Centre in Mumbai, aiming to assess genetic literacy among breast cancer patients in an urban setting. The analysis covers demographics, family history, cancer staging, tumor characteristics, hormone receptor status, and lifestyle factors.

TABLE 1. PATIENT DEMOGRAPHICS: AGE DISTRIBUTION

Age Group (Years)	*No. of Patients*	*Percentage*
Below 30	*2*	*1.6%*
30-40	*12*	*9.3%*
40-50	*37*	*28.7%*
50-60	*44*	*34.1%*
Above 60	*34*	*26.4%*

MENOPAUSAL STATUS:

- Postmenopausal: 103 patients (79.8%)
- Premenopausal: 38 patients (29.4%)
- Other (Hysterectomy/Perimenopausal/Unspecified): 12 patients

TABLE 2. FAMILY HISTORY OF CANCER: FAMILY CANCER HISTORY BREAKDOWN

Family History	*No. of Patients*	*Percentage*
Breast Cancer in Family	*18*	*14%*
Other Cancers in Family	*11*	*8.5%*
No Family History	*100*	*77.5%*

TABLE 3. BREAST CANCER STAGING & PROGRESSION: CANCER STAGE DISTRIBUTION

Stage	*No. of Patients*	*Percentage*
Stage I	*10*	*7.8%*
Stage II A	*32*	*24.8%*
Stage II B	*23*	*17.8%*
Stage III A	*13*	*10.1%*
Stage III B	*8*	*6.2%*
Stage III C	*3*	*2.3%*
Stage IV	*6*	*4.7%*
Unkonown / Missing	*34*	*26.3%*

TABLE 4. TUMOR CHARACTERISTICS: PATHOLOGY TYPES

Cancer Type	*No. of Patients*	*Percentage*
Ductal Carcinoma	*61*	*47.3%*
Mixed Ductal & Lobular	*16*	*12.4%*
Lobular Carcinoma	*7*	*5.4%*
DCIS (Ductal Carcinoma in Situ)	*3*	*2.3%*
Mucinous Carcinoma	*2*	*1.6%*
Cribriform Carcinoma	*1*	*0.8%*
Anaplastic Carcinoma	*1*	*0.8%*
Unknown/Missing	*38*	*29.4%*

TABLE 5. TUMOR SIZE DISTRIBUTION

Tumor Size (cm)	*No. of Patients*	*Percentage*
< 2 cm	*24*	*18.6%*
2 - 5 cm	*71*	*55.0%*
> 5 cm	*16*	*12.4%*
Unknown	*18*	*14.0%*

MENOPAUSAL STATUS:

Estrogen Receptor (ER) Status

- ER Positive: 77 patients (59.7%)
- ER Negative: 46 patients (35.7%)
- Unclear Data: 6 patients

Progesterone Receptor (PR) Status

- PR Positive: 59 patients (45.7%)
- PR Negative: 59 patients (45.7%)
- Unclear Data: 11 patients

HER2 (Human Epidermal Growth Factor Receptor 2) Status

- HER2 Positive (3+ or FISH Positive): 12 patients (9.3%)
- HER2 Negative (0, 1+, 2+ FISH Negative): 93 patients (72%)
- Unclear Data: 24 patients

LIFESTYLE & RISK FACTORS

TABLE 6. OBESITY & BMI (BODY MASS INDEX)

BMI Category	*No. of Patients*	*Percentage*
Underweight (<18.5 BMI)	*3*	*2.3%*
Normal Weight (18.5 - 24.9 BMI)	*27*	*20.9%*
Overweight (25 - 29.9 BMI)	*38*	*29.5%*
Obese (>30 BMI)	*49*	*38.0%*
Unknown	*12*	*9.3%*
BMI Category	*3*	*2.3%*

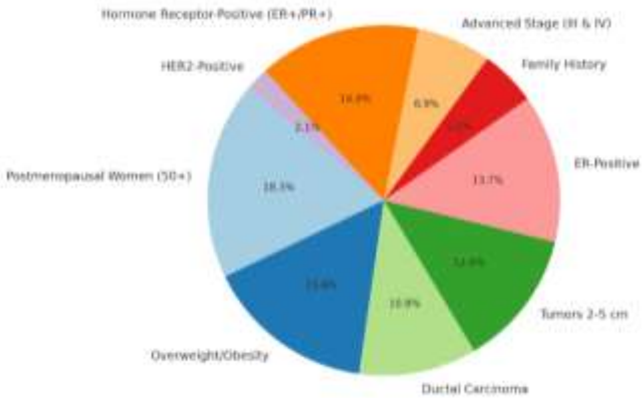

Fig 2. Breast Cancer Characteristics Distribution

METHODOLOGY

Proposed Method

1. Automated Risk Prediction: Users fill out a detailed questionnaire that pro vides information about their medical history and genetic background. The system then analyzes this data and classifies their genetic risk as low, moderate, or high.

2. Individualized Risk Evaluation: Based on the user's responses and the pre dicted risk level (low, moderate, high), personalized risk assessments are generated. This includes recommendations for further tests, lifestyle changes, and monitoring schedules.

3. Guidelines for Health: For each risk category, GeneGuard provides tailored health advice, such as food regimens, frequency of health screenings, and lifestyle modifications specific to the risk level.

4. User-Friendly Interface: The interface is designed to be simple and intuitive, making it easy for users to enter their information and receive feedback. Genetic risk and health advice are presented in a clear and understandable format.

5. Chatbot for Immediate Assistance: A built-in chatbot is available to answer users' questions in real-time, providing further explanations about genetic risks, the questionnaire, and recommendations. It serves as an accessible support system to enhance user engagement.

6. Modules for Education: Educational materials to increase knowledge about genetic risks and genetic counseling. articles and tutorials outlining the significance of early intervention and genetic testing.

Research Design :

Research design includes case studies, surveys, and interviews.

Research Approach:

GeneGuard uses a qualitative approach to gather data on the perspectives of genetic and non-genetic healthcare professionals. Additionally, quantitative data will be collected on the number of patients accessing genomic services and the time saved through GC integration.

Rationale: The qualitative approach helps in understanding the challenges and benefits of mainstreaming GCs, while the quantitative data provides measurable outcomes that can be evaluated for improvement in healthcare delivery.

Data Collection Methods :

Data collected through structured interviews with healthcare professionals and GCs. Surveys will also be used to gather broader quantitative data.

Instruments Used: The tools used for data collection include digital recorders for interviews, survey platforms like Google Forms for distributing questionnaires.

Field Visits and Data Sources:

Research involved several key site visits for data collection. Initially, we visited Kokilaben Hospital and the SDG Center in Vile Parle, where we gathered valuable insights into genetics. Following that, we conducted fieldwork at Bhaktivedanta Hospital in Matunga, collecting data personally. Lastly, we obtained additional data from the Oncology Center in Ghatkopar to ensure comprehensive research coverage.

Data Analysis Techniques:

Collected data will be analyzed using a combination of statistical methods and machine learning algorithms. Data processing and visualization will be performed using Pandas, Matplotlib, Cufflinks, and Plotly to clean, manipulate, and visualize genetic and clinical data. Scikit-learn will be used for algorithm-based predictions of genetic risk, as it is effective in pattern recognition within complex genetic datasets.

IMPLEMENTATION

Frontend (Flutter-based Client Interface):
1. UI/UX Design: Responsive design using Figma to create an intuitive and user-friendly interface.
2. Components: –
Login: Handles user authentication, secure login, and profile management.
3. Questionnaire Form: A form that collects input from the user, including medical history, symptoms, family health background, and lifestyle factors.
4. Results Page: Displays personalized risk predictions and recommendations based on the user's input data.

Backend:
1. User Management Module: Manages user registration, login, and profile management.
2. Questionnaire Processing: Handles inputs from the frontend, validates the data, and passes it to the prediction model.
3. Report Generation Module: Compiles results from the prediction model into a detailed, user-friendly report.

Prediction Model:
Data Flow: The model receives preprocessed input data from the backend and generates a prediction score for diseases.

Algorithms:
Uses decision trees, logistic regression, and transformer-based models (e.g., Gemini text-embedding-004) to predict cancer risks based on user inputs.

Chatbot

- PDF and DOCX Text Extraction: To handle unstructured data from documents, we used PDFMiner and PyMuPDF for extracting text from PDF files and docx2txt for Microsoft Word documents. These tools enable high-fidelity text extraction while preserving document structure.
- Recursive Character Text Splitting: Extracted documents were processed using recursive character-based text splitting to break down large text blocks into manageable and semantically coherent chunks. This technique ensures optimal context windows for downstream embedding and retrieval tasks.

- Transformer-based Text Embedding: We used Gemini text-embedding-004, a high-performance transformer-based model, to convert text chunks into dense vector representations. These embeddings capture semantic meaning and contextual relevance, forming the basis for similarity search.

- Approximate Nearest Neighbors (ANN): For efficient retrieval from a large corpus, we implemented Approximate Nearest Neighbor search using HNSW (Hierarchical Navigable Small World) graphs. This algorithm provides sub-linear search complexity while maintaining high recall accuracy.
- Retrieval-Augmented Generation (RAG): Retrieved top-k relevant text chunks are fed into the language model as additional context using a Retrieval-Augmented Generation framework. RAG enhances the factual grounding of responses and ensures dynamic access to external knowledge.

- Transformer-based Language Model: We integrated LLaMA 3 70B via **Groq**, leveraging its state-of-the-art generative capabilities for answering user queries in a natural, coherent, and context-aware manner.

- System and Human Message Templates: We used structured prompt engineering techniques to guide the language model. Prompts were composed of system messages (defining behavior and constraints) and human messages (user queries), ensuring deterministic and consistent outputs.

Machine Learning for Genetic Risk Prediction: To assess hereditary disease risks, we employ TensorFlow Keras and scikit-learn libraries.

- Train-Test-Validation Split
  We partitioned the dataset into training, validation, and testing subsets using train_test_split from sklearn.model_selection. This ensures unbiased evaluation and helps monitor the model's performance during and after training.

- Standardization
  Feature scaling was performed using StandardScaler from sklearn.preprocessing, which transforms the input features to have zero mean and unit variance. This standardization accelerates convergence and stabilizes the training process.
- One-Hot Encoding
  For categorical target labels, we applied to_categorical from tensorflow.keras.utils, converting integer labels into one-hot encoded vectors. This format is essential for multi-class classification using softmax activation.

We designed a multi-layer Deep Neural Network (DNN) using the Keras Sequential API, which includes the following components:

- Dense Layers: Fully connected layers that learn hierarchical feature representations.
- BatchNormalization: Applied after dense layers to normalize intermediate activations, improving training stability and speed.
- Dropout Layers: Introduced for regularization, reducing overfitting by randomly deactivating a fraction of neurons during training.
- Activation Functions:
  - ReLU (Rectified Linear Unit) for hidden layers, promoting non-linearity and efficient gradient propagation.
  - Softmax for the output layer, converting logits into probability distributions across multiple classes.

- Optimizer:
  We used the Adam optimizer, an adaptive learning rate optimization algorithm that combines the advantages of RMSProp and Momentum, providing fast convergence and stability.
- Loss Function:
  The model was trained using categorical cross-entropy, appropriate for multi-class classification problems with one-hot encoded labels.

- Accuracy:
  Model performance was primarily evaluated using accuracy, which measures the proportion of correctly classified instances over the total number of samples.

Key Advantages of Our Approach

By integrating these analytical tools, the study ensures:

- Accurate Genetic Risk Prediction – Machine learning models efficiently analyze genetic patterns.
- Improved Model Transparency – Visual tools provide intuitive explanations for genetic counselors.
- Optimized Model Performance – Hyperparameter tuning, such as selecting k in KNN, enhances prediction accuracy.
- Actionable Insights – Data-driven visualizations support informed decision-making in clinical settings.

OBSERVATIONS

- Most breast cancer patients (79.8%) are postmenopausal women aged 50 and above.
- Overweight and obesity (~68%) are strongly linked to breast cancer cases.
- Ductal carcinoma is the most common cancer type (47.3%).
- Tumors between 2-5 cm (55%) are most common, showing moderate early detection.
- ER-positive breast cancer is dominant (59.7%), making hormone therapy a primary treatment option.
- Family history plays a role in only 22.5% of cases, highlighting the significance of lifestyle and environmental factors.
- Early-stage detection is improving, but advanced cases (Stage III & IV) are still significant.
- Hormone receptor-positive (ER+/PR+) breast cancer is more prevalent, meaning many patients can benefit from hormone therapy.
- HER2-positive cases are relatively low (9.3%), indicating fewer patients require targeted HER2 therapy.

GENETIC TESTING ASSESSMENT BASED ON OUR BREAST CANCER POPULATION DATA FINDINGS

Genetic testing plays a crucial role in evaluating breast cancer risk, guiding treatment decisions, and improving early detection. Our analysis of population data highlights key areas where genetic testing is particularly relevant:

1. Identifying Hereditary Risk in Patients with a Family History (22.5%)

- Our data shows that 22.5% of patients reported a family history of cancer. Those with close relatives affected by breast or ovarian cancer may carry high-risk mutations in genes such as BRCA1, BRCA2, TP53, PTEN, and PALB2.
- Genetic testing can confirm inherited mutations, allowing for early intervention through increased screening, risk-reducing surgeries, or lifestyle modifications.

2. Personalized Treatment Based on Hormone Receptor and HER2 Status

- ER-Positive (59.7%) & PR-Positive (45.7%) Patients:
    - Genetic testing can detect ESR1 and other hormone-related mutations that may influence resistance to endocrine therapy.
- HER2-Positive (9.3%) Patients:
    - Genetic profiling helps confirm HER2 amplification, guiding treatment with targeted therapies like trastuzumab (Herceptin).
- Triple-Negative Breast Cancer (ER-/PR-/HER2-):
    - These patients are more likely to carry BRCA mutations, making them candidates for PARP inhibitors or clinical trials exploring precision therapies.

3. Early Detection and Prevention for High-Risk Individuals

- With 38% of patients classified as obese, a known risk factor for breast cancer, genetic testing can help assess risk in high-BMI individuals with a family history.
- High-risk gene carriers (BRCA1/2) may opt for:
    - Enhanced screening (early and more frequent mammograms, MRI scans).
    - Preventive mastectomy to reduce cancer risk.
    - Lifestyle modifications to address non-genetic risk factors such as obesity and hormonal imbalances.

4. Predicting Recurrence and Metastasis Risk

- Genetic tests like Oncotype DX, MammaPrint, and Prosigna are valuable in assessing recurrence risk for early-stage patients.
- Among patients with advanced-stage tumors (Stage III & IV, 23.3%), next-generation sequencing (NGS) can identify mutations that may be targeted with novel drug therapies.

5. Expanding Research on Non-Familial Breast Cancer Cases

- Our data reveals that 77.5% of cases lack a family history, emphasizing the role of sporadic and environmental mutations in breast cancer development.
- Genetic testing can help detect somatic mutations (e.g., PIK3CA, TP53, AKT1) that influence tumor progression and response to treatment.
- These insights contribute to precision medicine, ensuring treatments are tailored based on individual genetic profiles rather than a one-size-fits-all approach.

Based on the dataset, genetic testing should be prioritized for:

- Patients with a family history of breast/ovarian cancer.
- Patients with triple-negative breast cancer (higher likelihood of BRCA mutations).
- Patients diagnosed at a younger age (<40 years old), where hereditary factors may play a role.
- High-risk individuals (obese, postmenopausal, or those with large tumors).
- Patients in advanced stages (Stage III & IV) for identifying targetable mutations.

This genetic testing assessment, derived from our population data findings, underscores its critical role in breast cancer risk evaluation, early detection, and personalized treatment. By integrating genetic insights into clinical practice, we can enhance patient outcomes and advance breast cancer research.

The findings suggest a potential gap in genetic literacy among patients at the hospital.

Here’s why:

Low Awareness of Family History & Hereditary Risk (22.5%): Only 22.5% of patients reported a family history of cancer, despite the well-documented role of hereditary mutations (BRCA1, BRCA2, TP53, etc.) in breast cancer. This could indicate that patients were not fully aware of the importance of family history in cancer risk assessment.

Limited Utilization of Genetic Testing: While genetic testing can inform treatment (e.g., HER2-targeted therapies, PARP inhibitors), there’s no mention of widespread testing among patients. If genetic testing was underutilized, it suggests that patients may not have been adequately informed about its benefits or eligibility criteria.

High Percentage of Non-Familial Cases (77.5%): The fact that 77.5% of cases had no reported family history could mean that genetic education was not emphasized, leading patients to underestimate the role of genetic and somatic mutations in cancer development. Many non-familial cases still have genetic components (e.g., PIK3CA, TP53 mutations), but patients may not have been educated on sporadic vs. inherited mutations.

Lifestyle & Risk Awareness (38% Obesity Rate): With 38% of patients classified as obese, and obesity being a known risk factor for breast cancer, genetic literacy could help patients understand gene-environment interactions. If education on modifiable vs. non-modifiable genetic risk factors was lacking, patients may not have been equipped to make informed lifestyle choices.

Our Recommendation on how to improve Genetic Literacy and providing Genetic Education in Healthcare Systems in India

“Genetic counseling is essential for bridging the gap between genetic discoveries and patient understanding. Without it, genetic information remains underutilized in clinical settings” [32].

Genetic literacy is crucial for enabling individuals to understand genetic risk factors, make informed medical decisions, and adopt preventive healthcare measures. In India, where genetic disorders and hereditary diseases like breast cancer, thalassemia, sickle cell anemia, and congenital disabilities are prevalent, improving genetic education in the healthcare system is essential.

Integrating Genetic Counseling into Healthcare Services: Genetic counseling helps individuals understand their genetic risks and make informed health decisions. In India, genetic counseling is not yet widely accessible, leading to misconceptions and underutilization of genetic testing.

Suggested Strategies:

- Mandatory Genetic Counseling in Hospitals: Large hospitals should have dedicated genetic counselors to guide patients undergoing genetic testing.
- Training Healthcare Professionals: Doctors and nurses should receive basic genetic literacy training to communicate test results effectively.
- Pre-Test and Post-Test Counseling: Counseling should be provided before and after genetic tests to ensure patients understand the implications of their results.

Incorporating Genetics into Medical and Nursing Curricula: Currently, genetics education in Indian medical schools is minimal, and many doctors are not trained to interpret genetic test results.

Suggested Strategies

- Expanding Genetics Education in Medical Schools: Introduce comprehensive courses on medical genetics, genomics, and bioinformatics.
- Continuing Medical Education (CME) Programs: Regular training workshops for doctors, nurses, and paramedics on recent advances in genetics.
- Collaboration with Global Institutes: Partnerships with international genetics institutions for faculty exchange and research.

Public Awareness Campaigns on Genetic Literacy: A lack of awareness about genetic disorders results in low uptake of genetic testing. Many Indians perceive genetic diseases as fate-based rather than scientifically preventable.

Suggested Strategies:

- Mass Media Campaigns: Government and NGOs should use television, radio, and social media to spread awareness.
- School & College Awareness Programs: Introduce basic genetics education in school curricula.
- Community Workshops: Conduct regional genetic literacy programs in rural areas to explain genetic conditions and testing benefits.

Making Genetic Testing Affordable and Accessible: Genetic tests in India are often expensive and limited to urban hospitals, making them inaccessible to lower-income populations.

Suggested Strategies:

- Subsidized Genetic Testing Programs: The government should introduce free or low-cost genetic testing for high-risk populations.
- Expansion of Genetic Labs to Rural Areas: Establish regional genetic testing centers with telemedicine support.
- Insurance Coverage for Genetic Tests: Advocacy for health insurance policies to cover genetic testing and counseling.

Addressing Ethical and Cultural Barriers to Genetic Testing: Cultural beliefs, fear of stigma, and ethical concerns about genetic discrimination hinder genetic literacy and testing.

Suggested Strategies:

- Confidentiality Policies: Enforce strict data protection laws to prevent genetic discrimination in employment and insurance.
- Culturally Sensitive Genetic Education: Genetic literacy programs should address religious and cultural concerns about testing.
- Involving Religious and Community Leaders: Local leaders should advocate for genetic health awareness to reduce stigma.

Conclusion

Our findings indicate that genetic literacy in India remains insufficient, particularly in clinical settings where patients lack adequate guidance on genetic testing. Despite the growing significance of genetic testing in diagnosing hereditary conditions and guiding precision medicine, gaps in education, accessibility, and awareness persist. Patients at high genetic risk often do not receive the necessary information to make informed health decisions, suggesting that hospitals and healthcare providers must improve genetic education and counseling services.

By implementing our recommendations, India can bridge the gap between genetic advancements and patient awareness, ultimately leading to better disease prevention, personalized treatment, and improved public health outcomes. A multi-disciplinary, patient-centered approach is essential for integrating genetic literacy into India’s healthcare system and ensuring that individuals can make informed decisions regarding their genetic health.

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AUTHOR INFORMATION

AUTHORS AND AFFILIATIONS:

Usha Mittal Institute of Technology (UMIT), SNDT Women's University
Prerna Jha, Harshita Singh, Harshada Jagade, Shraddha Karande

Databyte Systems and Software:
Mr. Vishram Bapat

CONTRIBUTION

Prerna Jha, Harshita Singh, and Harshada Jagade established this project in 2024 as an academic major project. Prerna Jha and Harshada Jagade collected the data through various hospitals. Harshita Singh analyzed the data and formatted it.
Prof. Shraddha Karande conducted feedback session along with Mr. Vishram Bapat. All authors contributed to editing the paper and approving the final manuscript.

CORRESPONDING AUTHORS

Correspondance to Prerna Jha.

ETHICS DECLARATIONS

Usha Mittal Institute of Technology, SNDT, approved each phase of the project. We, the authors of this manuscript, confirm that the submitted work is original, has not been published previously, and is not under consideration for publication elsewhere.
We further declare that this study adheres to the ethical standards of scientific research and complies with the ethical publication practices outlined by the Committee on Publication Ethics (COPE).
Ethical considerations in this study include ensuring informed consent, protecting participant confidentiality, and maintaining transparency in data collection processes. Participants were fully informed about the nature and purpose of the research and voluntarily agreed to participate. Their personal and medical data is anonymized to protect privacy, and all sensitive information is securely stored.
All authors have read and approved the final manuscript and agree with its submission to the *Journal of Community Genetics*.

COMPETING INTERESTS

The authors declare no competing interests.

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