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Maternal awareness and misconceptions about genetic disorders: a cross-sectional study among mothers of affected children at PIMS Hospital, Islamabad

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Abstract

Genetic disorders impose a considerable health burden in low- and middle-income countries, where high consanguinity rates, limited access to services, and low health literacy intensify their impact. This study assessed awareness, knowledge, and misconceptions about genetic disorders among mothers of affected children at the Pakistan Institute of Medical Sciences (PIMS), Islamabad, and examined associated sociodemographic factors. A cross-sectional survey was conducted from April to May 2025 among 100 mothers of children with confirmed genetic disorders via a structured, interviewer-administered questionnaire. The mean participant age was 31.13 ± 6.06 years; 36% were illiterate, and 52% resided in urban areas. While 78% had heard of genetic disorders, only 18% knew of local genetic testing or counseling services, and 24% had received information from healthcare providers. Good awareness was seen in 16% and poor awareness in 78% of the participants; good knowledge was found in 25%, whereas 66% had poor knowledge. Misconceptions persisted, with 21% attributing genetic disorders to bad luck or punishment and 16% believing they were contagious; 38% demonstrated good understanding, and 46% demonstrated poor understanding. Education and socioeconomic class were significantly associated with awareness ($p = 0.011$, $p = 0.001$), knowledge ($p < 0.001$, $p = 0.002$), and beliefs ($p < 0.001$, $p < 0.001$), whereas age and residence were not significantly associated. The findings highlight a gap between general awareness and functional understanding, emphasizing the need for targeted, literacy-sensitive interventions to improve maternal genetic health literacy and support informed healthcare decision-making.

Keywords

Genetic diseases, inborn; Health knowledge, attitudes, practice; Mothers; Socioeconomic factors; Consanguinity

1. Introduction

Genetic disorders contribute significantly to the global burden of disease, affecting millions of children and imposing considerable emotional, medical, and financial strain on families and healthcare systems. Recent estimates suggest that approximately 6% of newborns worldwide are born with serious birth defects, including congenital disorders with structural or functional anomalies [1]. The Global Burden of Disease Study 2021 highlights persistent disparities in disability-adjusted life years (DALYs) attributable to

congenital anomalies, particularly in low-resource settings where access to preventive and diagnostic services remains limited [2]. Global modeling indicates that chromosomal abnormalities and single-gene disorders—together accounting for approximately 8.9% of congenital anomalies—exhibit substantial variation in prevalence between regions, influenced by factors such as maternal age, rates of consanguinity, environmental exposures, and healthcare access [3].

In Pakistan, a low- and middle-income country (LMIC), the burden of genetic disorders is intensified by socioeconomic inequities, high rates of consanguinity, and limited genetic counseling infrastructure [4,5]. Nationally, nearly two-thirds of marriages occur between blood relatives—most commonly first or second cousins—substantially increasing the risk of autosomal recessive disorders [6]. For example, a recent study in Balochistan reported consanguineous unions in half of all congenital anomaly cases, with neurological and limb defects among the most common presentations [5]. The evidence indicates that consanguinity increases the frequency of autosomal recessive disorders by up to twofold compared with nonconsanguineous couples [7].

Maternal awareness plays a pivotal role in early recognition, health-seeking behavior, and the management of genetic disorders. However, in LMICs, awareness levels are often limited due to educational, cultural, and informational barriers. Studies in Pakistan indicate that misconceptions about the causes of genetic disorders are widespread, with a substantial proportion of the population attributing them to infections, nutritional factors, or supernatural beliefs, and awareness of the role of consanguinity in increasing genetic risk is significantly greater among educated individuals than among those with little or no formal schooling [4,8,9]. Comparable patterns have been observed globally. In India, only 36.5% of pregnant women demonstrated an understanding of prenatal genetic screening [10], whereas in Rwanda, 53.8% of women could not name a single genetic condition, although many expressed a willingness to undergo carrier testing if available [11].

Low maternal literacy not only hinders understanding but also delays diagnosis and treatment. A 2022 cohort study linked limited schooling with reduced healthcare utilization for genetic conditions [12]. Misconceptions rooted in cultural beliefs—such as attributing disorders to spiritual punishment or the "evil eye"—further complicate clinical management [8,13]. In Pakistan, diagnostic delays often extend beyond four years, with many families resorting to spiritual healers or discontinuing biomedical treatments when symptoms appear to improve [14,15].

This study seeks to address this gap by evaluating awareness, beliefs, and misconceptions about genetic disorders among mothers of affected children at the Pakistan Institute of Medical Sciences (PIMS) Hospital in Islamabad. The primary objective of this study was to assess the level of awareness among mothers regarding genetic disorders, specifically among those whose children are admitted with such conditions at PIMS Hospital. The study further aims to explore the prevalent misconceptions and myths held by these mothers about the nature, transmission, and management of genetic disorders. Additionally, the research seeks to examine how key demographic factors—particularly maternal age, literacy status, locality, and socioeconomic class—influence both awareness levels and the presence of misconceptions. By identifying these knowledge gaps and demographic patterns, this study aims to provide insights that can guide targeted educational interventions and public health strategies aimed at improving maternal understanding of genetic health issues.

2. Methods

2.1. Study design, setting and duration

This cross-sectional study was conducted at PIMS Hospital, Islamabad, between April and May 2025. The study population included mothers of children diagnosed with genetic disorders who were admitted to the hospital during the study period.

2.2. Ethics consideration

Ethical approval for the study was obtained from the Ethical Research Review Committee (ERRC) of the Islamabad Hospital, PIMS (No. F-5-2/2024(ERRC)/PIMS). In addition, written permission was obtained from the PIMS Hospital administration to conduct data collection within the hospital premises.

2.3. Sample size and sampling method

The sample size was calculated via the single population proportion formula, considering a prevalence rate of 5.9% for genetic disorders, as reported in previous literature, with a 95% confidence level and 5% margin of error, resulting in a minimum required sample size of approximately 86 [16]. To account for potential dropouts, the sample size was increased by 25%, resulting in a final target of 108 participants, who were approached via a purposive sampling technique.

2.4. Selection criteria

Mothers were eligible for inclusion if they had a child with a confirmed diagnosis of a genetic disorder admitted to PIMS Hospital during the study period, were aged 18 years or older, and were willing to provide written informed consent. Mothers who were not the primary caregiver, had severe communication difficulties, or declined to participate were excluded from the study.

2.5. Questionnaire development

A structured, interviewer-administered questionnaire was developed to assess maternal awareness, knowledge, and misconceptions regarding genetic disorders. The tool was adapted from previously validated instruments used in studies assessing genetic literacy and maternal perceptions in LMICs [11,17,18,19,20], with minor modifications to ensure cultural appropriateness and contextual relevance. The questionnaire comprises four sections, including demographic information (age, education, residence, and socioeconomic class); awareness of genetic disorders (three binary yes/no items: having heard of genetic disorders, knowledge of genetic counseling/testing centers in Pakistan, and receiving information from a healthcare provider); and knowledge-based questions (three statements covering prevention through medical interventions, inheritance patterns, and perceived causes; responses: true/false/don't know); and beliefs and misconceptions (eight culturally relevant statements addressing common myths and misconceptions about genetic disorders; responses: true/false/don't know). The instrument was pretested on five mothers of children with confirmed genetic disorders at the same facility to ensure clarity, linguistic suitability, and cultural sensitivity, with minor revisions made before final administration.

2.6. Data collection

Data were collected through face–to-face interviews conducted by a trained female research assistant fluent in Urdu and local languages, held in a private area within PIMS

Hospital, Islamabad, to ensure confidentiality and participant comfort, with each interview lasting approximately 10–15 minutes. Responses were recorded on printed questionnaires and subsequently entered into a secure database for analysis. Written informed consent was obtained from all participants prior to data collection.

2.7. Study measures

The awareness score was derived from three binary (yes/no) items, with one point awarded for each "yes" response (range: 0–3). Scores were classified as good (3 points), moderate (2 points), or poor (0–1 points) [21]. Similarly, the knowledge score was calculated from the three knowledge-based questions, assigning one point for each correct response, with the same classification applied for good (3 points), moderate (2 points), and poor (0–1 points) answers. The misconception score was calculated from the eight belief/misconception items, with one point awarded for each correct response, resulting in a possible range of 0–8 points. For misconceptions, higher scores indicate better understanding and are categorized as low misconceptions (0–4 points), moderate misconceptions (5–6 points), and high misconceptions (7–8 points), with predefined score ranges applied in awareness research [22]. For analysis, age (continuous) was transformed into two categories, ≤30 years and >30 years, reflecting younger and older maternal age groups, respectively. Education was dichotomized into "literate" (able to read/write at any formal education level) and "illiterate" (no formal education).

2.8. Statistical analysis

The data were entered and analyzed via IBM SPSS Statistics (version 27.00, IBM Corp., Armonk, NY, USA). Descriptive statistics were used to summarize participant characteristics, awareness, knowledge, and misconceptions regarding genetic disorders, with categorical variables presented as frequencies and percentages. The associations between sociodemographic variables and outcome variables, including awareness level, knowledge level, and misconception level, were assessed via the chi-square test. A p value of < 0.05 was considered to indicate statistical significance.

3. Results

Table 1 shows that the mean age of the participants was 31.13 years (SD ± 6.06). Over one-third of the mothers (36%) were illiterate, while 13% had completed graduate-level education or higher. The participants were almost evenly distributed between urban (52%) and rural (48%) areas. With respect to socioeconomic class, both the working and middle classes accounted for 43% each, with 14% belonging to the upper class.

Table 1. Sociodemographic characteristics of the study participants (N = 100).

Variables		Frequency (%)	Mean ± S.D.
Age (in years)		-	31.13 ± 6.06
Education	Illiterate	36 (36.00)	-
	Primary	11 (11.00)	-
	Secondary	18 (18.00)	-
	Higher secondary	22 (22.00)	-
	Graduate and above	13 (13.00)	-
Residence	Urban	52 (52.00)	-
	Rural	48 (48.00)	-
Socioeconomic class	Working class	43 (43.00)	-
	Middle class	43 (43.00)	-

Variables		Frequency (%)	Mean ± S.D.
	Upper class	14 (14.00)	-

Table 2 shows that most participants (78%) reported awareness of genetic disorders; however, only 18% knew about local centers offering genetic testing or counseling, and 24% had received information from healthcare providers. Overall, 16% of mothers demonstrated good awareness, whereas 78% had poor awareness. With respect to knowledge, 44% believed that genetic disorders could be prevented through medical interventions, and 51% thought that they were always inherited. One-fourth (25%) of the participants had good knowledge, whereas 66% had poor knowledge. In terms of beliefs and misconceptions, 62% recognized that cousin marriage increases the risk of genetic disorders, whereas 51% believed that these conditions are always apparent at birth. Misconceptions such as attributing genetic disorders to bad luck or punishment (21%) or considering them contagious (16%) were also noted. Overall, 38% demonstrated good understanding, 16% had moderate misconceptions, and 46% had poor understanding.

Table 2. Awareness, knowledge, and misconceptions about genetic disorders among the study participants (N = 100).

Variables		Frequency (%)
Level of Awareness		
Awareness of genetic disorders	Yes	78 (78.00)
	No	22 (22.00)
Knowledge of local genetic testing/counseling services	Yes	18 (18.00)
	No	82 (82.00)
Information received from healthcare providers	Yes	24 (24.00)
	No	76 (76.00)
Awareness level	Good awareness	16 (16.00)
	Moderate awareness	6 (6.00)
	Poor awareness	78 (78.00)
Level of Knowledge		
Perceived preventability of genetic disorders through medical interventions	True	44 (44.00)
	False	20 (20.00)
	Don't know	36 (36.00)
Belief that genetic disorders are always inherited	True	51 (51.00)
	False	27 (27.00)
	Don't know	22 (22.00)
Belief that maternal lifestyle or diet can cause genetic disorders	True	35 (35.00)
	False	32 (32.00)
	Don't know	32 (32.00)
Knowledge level	Good knowledge	25 (25.00)
	Moderate knowledge	9 (9.00)
	Poor knowledge	66 (66.00)
Beliefs and Misconceptions Level		
Belief that genetic disorders are contagious	True	16 (16.00)
	False	44 (44.00)
	Don't know	40 (40.00)
Belief that genetic disorders are always apparent at birth	True	51 (51.00)
	False	17 (17.00)
	Don't know	32 (32.00)
Belief that cousin marriage increases genetic disorder risk	True	62 (62.00)
	False	26 (26.00)

Variables		Frequency (%)
	Don't know	12 (12.00)
Belief that no intervention is possible after diagnosis	True	30 (30.00)
	False	45 (45.00)
	Don't know	25 (25.00)
Belief that genetic disorders result from bad luck or punishment	True	21 (21.00)
	False	49 (49.00)
	Don't know	30 (30.00)
Belief that genetic testing is only useful postnatally	True	38 (38.00)
	False	43 (43.00)
	Don't know	19 (19.00)
Belief that healthy-looking parents cannot have a child with a genetic disorder	True	10 (10.00)
	False	79 (79.00)
	Don't know	11 (11.00)
Belief that a healthy first child guarantees subsequent healthy children	True	28 (28.00)
	False	48 (48.00)
	Don't know	24 (24.00)
Beliefs and misconceptions level	Good understanding	38 (38.00)
	Moderate misconceptions	16 (16.00)
	Poor understanding	46 (46.00)

Table 3 shows the associations between sociodemographic characteristics and awareness levels regarding genetic disorders. Education and socioeconomic class were significantly associated with awareness ($p = 0.011$ and $p = 0.001$, respectively), with higher awareness observed among literate participants and those from the middle or upper socioeconomic classes. Age and place of residence were not significantly associated with awareness level.

Table 3. Associations between sociodemographic characteristics and level of awareness of genetic disorders among the study participants (N = 100).

Variables		Level of Awareness			*p* Value *
		Good Awareness (n = 16)	Moderate Awareness (n = 6)	Poor Awareness (n = 78)	
		N (%)	N (%)	N (%)	
Age	> 30 years	7 (7.00)	2 (2.00)	45 (45.00)	0.344
	≤ 30 years	9 (9.00)	4 (4.00)	33 (33.00)	
Education	Literate	15 (15.00)	2 (2.00)	47 (47.00)	0.011 *
	Illiterate	1 (1.00)	4 (4.00)	31 (31.00)	
Residence	Urban	9 (9.00)	3 (3.00)	40 (40.00)	0.932
	Rural	7 (7.00)	3 (3.00)	38 (38.00)	
Socioeconomic class	Working class	0 (0.00)	3 (3.00)	40 (40.00)	0.001 *
	Middle class	10 (10.00)	3 (3.00)	30 (30.00)	
	Upper class	6 (6.00)	0 (0.00)	8 (8.00)	

* Data were analyzed by using the chi-square test. ** Significant value ($p < 0.05$).

Table 4 presents the associations between sociodemographic characteristics and knowledge levels. Education ($p < 0.001$) and socioeconomic class ($p = 0.002$) were significantly associated with knowledge, with better knowledge observed among literate participants and those from higher socioeconomic classes. Age and residence were not significantly related to knowledge level.

Table 4. Associations between sociodemographic characteristics and level of knowledge regarding genetic disorders among the study participants (N = 100).

Variables		Level of Knowledge			*p* Value *
		Good Knowledge (n = 25)	Moderate Knowledge (n = 9)	Poor Knowledge (n = 66)	
		N (%)	N (%)	N (%)	
Age	> 30 years	15 (15.00)	3 (3.00)	36 (36.00)	0.383
	≤ 30 years	10 (10.00)	6 (6.00)	30 (30.00)	
Education	Literate	23 (23.00)	9 (9.00)	32 (32.00)	< 0.001 *
	Illiterate	2 (2.00)	0 (0.00)	34 (34.00)	
Residence	Urban	17 (17.00)	6 (6.00)	29 (29.00)	0.080
	Rural	8 (8.00)	3 (3.00)	37 (37.00)	
Socioeconomic class	Working class	6 (6.00)	0 (0.00)	37 (37.00)	0.002 *
	Middle class	13 (13.00)	6 (6.00)	24 (24.00)	
	Upper class	6 (6.00)	3 (3.00)	5 (5.00)	

* Data were analyzed by using the chi-square test. ** Significant value ($p < 0.05$).

Table 5 shows the associations between sociodemographic characteristics and beliefs/misconceptions about genetic disorders. Education ($p < 0.001$) and socioeconomic class ($p < 0.001$) were significantly associated with beliefs and misconceptions, with accurate beliefs and fewer misconceptions being more common among literate participants and those in higher socioeconomic classes. No significant associations were found for age or residence.

Table 5. Associations between sociodemographic characteristics and the level of beliefs and misconceptions about genetic disorders among study participants (N = 100).

Variables		Belief and Misconceptions Level			*p* Value *
		Accurate Beliefs/Low Misconceptions (Good Understanding) (n = 38)	Moderate Misconceptions (n = 16)	High Misconceptions (Poor Understanding) (n = 46)	
		N (%)	N (%)	N (%)	
Age	> 30 years	22 (22.00)	11 (11.00)	21 (21.00)	0.232
	≤ 30 years	16 (16.00)	5 (5.00)	25 (25.00)	
Education	Literate	33 (33.00)	11 (11.00)	20 (20.00)	< 0.001 *
	Illiterate	5 (5.00)	5 (5.00)	26 (26.00)	
Residence	Urban	24 (24.00)	7 (7.00)	21 (21.00)	0.215
	Rural	14 (14.00)	9 (9.00)	25 (25.00)	
Socioeconomic class	Working class	9 (9.00)	6 (6.00)	28 (28.00)	< 0.001 *
	Middle class	21 (21.00)	5 (5.00)	17 (17.00)	
	Upper class	8 (8.00)	5 (5.00)	1 (1.00)	

* Data were analyzed by using the chi-square test. ** Significant value ($p < 0.05$).

4. Discussion

The findings of this study indicate that while a majority of mothers were aware of genetic disorders in general terms, detailed knowledge about their causes, prevention, and available healthcare services was limited, and misconceptions remained prevalent. Educational attainment and socioeconomic status emerged as key factors influencing

awareness, knowledge, and the accuracy of beliefs, whereas age and place of residence appeared to have little impact. The persistence of misconceptions alongside limited awareness of local genetic services suggests a gap between general familiarity with the concept of genetic disorders and a functional understanding that could support informed decision-making and health-seeking behavior.

Our findings align with previous research in Pakistan and other LMICs showing that general awareness of genetic disorders may be relatively common, but detailed knowledge and understanding remain poor [22,23,24,25]. Similar to a Pakistan-based study, where almost 80% of respondents were familiar with the term genetic disorders and 39.3% had heard about genetic testing before the survey, our participants demonstrated high general awareness but limited familiarity with specific services and preventive strategies [22]. A similar trend was observed in an Indian study, where only 36.5% of pregnant women knew about prenatal genetic screening tests, indicating that general awareness often does not translate into a deeper understanding [10]. The persistence of misconceptions—such as the belief that genetic disorders are always inherited or always visible at birth—mirrors findings from Rwanda, where more than half of the women surveyed could not name a single genetic condition despite having heard of the term [11]. These similarities may reflect shared barriers such as low genetic literacy, limited access to counseling, and inadequate integration of genetic health topics into primary care.

The significant associations between higher educational attainment and improved awareness, knowledge, and reduced misconceptions observed in this study align with evidence from various settings. Previous research in Pakistan and other LMICs has consistently shown that literacy equips mothers with the capacity to critically assess health information, leading to more accurate perceptions of genetic disorders [12,16]. Similarly, international studies have indicated that increased years of formal education are often linked to higher genetic literacy and a greater ability to reject misconceptions [26,27,28]. The association between socioeconomic class and better awareness in our findings is also in line with global evidence suggesting that households with greater economic resources are more likely to afford diagnostic services, seek specialist consultations, and participate in health education programs [14,29]. This relationship may be explained by better access to reliable health information, stronger health-seeking behaviors, and more frequent interactions with healthcare providers among educated and higher-income families [30,31].

In contrast, our finding that maternal age and place of residence did not significantly influence awareness, knowledge, or beliefs differs from patterns reported in several earlier studies. Research in certain regions of LMICs has suggested that rural mothers often demonstrate lower awareness of genetic risk factors than their urban counterparts do, whereas studies in other LMICs, such as Rwanda, have also documented rural–urban knowledge gaps [17,32]. The absence of such disparities in our study may be attributable to our hospital-based sampling at a tertiary care facility, where both urban and rural participants had already navigated healthcare referral pathways and may have had prior exposure to genetic health information.

The lack of a significant association between maternal age and genetic health literacy in our results also contrasts with findings from settings where older mothers were expected to possess greater knowledge owing to accumulated life experience [33]. In our context, the absence of an age effect may suggest that experience alone is insufficient to improve understanding without targeted health education [34]. This interpretation is consistent with studies indicating that formal education and structured information delivery are more influential determinants of knowledge than age is [35].

A key strength of our study lies in its focus on mothers of children with confirmed genetic disorders, ensuring that responses were grounded in lived experience rather than hypothetical scenarios. The use of a culturally adapted, pretested questionnaire improved the reliability of the responses in the local context. However, several limitations should be noted. The purposive sampling method and single-institution setting may limit generalizability to broader maternal populations in Pakistan, especially those without access to tertiary care. The cross-sectional design precludes the assessment of causal relationships between sociodemographic variables and knowledge or beliefs. Self-reported awareness and knowledge are also subject to recall and social desirability biases, which may lead to overestimation of understanding. Despite these limitations, this study provides valuable insights into the gaps in maternal genetic literacy in high-risk populations.

5. Conclusions

This study highlights that while a majority of mothers of children with genetic disorders possessed general awareness of these conditions, their detailed knowledge and accurate understanding were notably limited, with several misconceptions still prevalent. Educational attainment and socioeconomic status emerged as significant determinants of awareness, knowledge, and belief accuracy, emphasizing the role of social and educational factors in shaping genetic health literacy. The gap between basic awareness and actionable understanding signals a critical barrier to timely diagnosis, appropriate healthcare utilization, and informed decision-making in the management and prevention of genetic disorders.

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Consent to participate: Not applicable.

Data availability: The data supporting this study's findings are available from the corresponding author, Areesha Rashid, upon reasonable request.

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