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Community Genetics Perspectives on Reproductive Decision-Making: Knowledge, Attitudes, and Barriers to Pre-implantation Genetic Testing Acceptance Among Families Affected by Hemoglobinopathies in Oman

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Abstract

Background

Sickle cell disease (SCD) and beta-thalassemia major (βTM) are common genetic disorders in Oman. Pre-implantation genetic testing (PGT) enables selection of unaffected and human leukocyte antigen (HLA)-matched embryos, offering an alternative reproductive option for affected families. However, PGT utilization remains limited among at-risk populations.

Methods

A questionnaire-based survey was conducted among parents of children with SCD or βTM attending the Royal Hospital, Muscat, Oman. A total of 385 participants were recruited and interviewed. Data were analyzed using SPSS (Statistical Package for the Social Sciences) version 23, and associations between categorical variables were assessed using Chi-squared tests.

Results

Most participants (58.2%) were unaware of PGT, and 92.2% had not previously undergone the procedure. Nevertheless, 86.4% expressed willingness to consider PGT in the future. Prevention of disease transmission was identified as the main advantage by 57.7% of participants. Higher education and income were significantly associated with greater awareness of PGT ($p < 0.001$ and $p = 0.007$, respectively). Gender, education level, income, and prior PGT knowledge were associated with having undergone the procedure ($p < 0.05$). Participants who had undergone premarital screening demonstrated significantly greater awareness of genetic risk ($p < 0.001$).

Conclusions

Despite limited awareness, acceptance of PGT among parents of children with SCD and βTM was high. This study reveals substantial gaps between knowledge and willingness to pursue PGT. Targeted education, genetic counseling integration, and accessible financial support are essential to address identified barriers and promote informed reproductive decision-making in affected families.

INTRODUCTION

Sickle cell disease (SCD) and beta-thalassemia major (βTM) are prevalent genetic disorders (Mangla et al. 2024; Origa 2017). According to the World Health Organization (WHO), these diseases impact up to 5% of the global population, posing a significant public health concern, particularly in regions such as the Mediterranean and the Middle East (World Health Organization 2006). Thalassemia syndromes and related hemoglobinopathies are the most common group of monogenic disorders worldwide, with

approximately 7% of the global population being carriers (Kohne 2011). The WHO reports that approximately 300,000 infants are born annually with thalassemia syndromes or sickle cell anemia. Although the percentage of thalassemia carriers globally is higher than that of sickle cell carriers, the higher frequency of the sickle cell gene in certain regions results in a greater number of affected births (World Health Organization 2006).

It is essential for genetic counsellors to be knowledgeable regarding the reproductive options available for carriers of genetic diseases. In Oman, the prevalence of SCD ranges from 0.2% to 0.3%, while homozygous βTM occurs in approximately 0.07% to 0.08% of the population. In turn, the prevalence of the sickle cell trait and β-thalassemia trait is 4.8–6% and 2.6%, respectively (Al-Riyami et al. 2001; Al-Riyami and Ebrahim 2003). These rates are significant, making Oman one of the Arab Gulf states with the highest prevalence of SCD (Al-Mashaikhi et al. 2022). Unfortunately, experts have observed even higher rates of children diagnosed with hemoglobinopathies in hematology clinics. Moreover, it is crucial to note that Oman currently lacks a national registry for hemoglobinopathies. In 1999, Oman introduced a national premarital screening and counseling program aimed at reducing the incidence of congenital anomalies and inherited hemoglobin disorders (AlKamyani and Al Hinai 2018).

Both SCD and βTM are characterized by abnormalities in the beta-globin component of hemoglobin (Mangla et al. 2024; Origa 2017). Specifically, SCD arises from a genetic mutation causing a substitution of hydrophobic valine for hydrophilic glutamic acid at position 6 on chromosome 11 (Higgs and Wood 2008). This leads to the formation of abnormal hemoglobin S, which causes red blood cells to become rigid and sickle-shaped. The impact on affected individuals is profound, often leading to chronic hemolytic anemia, pain crises, and significant organ damage, with symptoms typically manifesting after infancy as fetal hemoglobin levels decrease (Mangla et al. 2024). In contrast, βTM is caused by a deficiency in the production of beta-globin chains due to various point mutations in the beta-globin gene on chromosome 11 (Muncie Jr and Campbell 2009). Due to the complete or near-complete absence of beta-globin chains, patients often exhibit severe anemia and require regular blood transfusions and iron chelation therapy to manage their condition (Origa 2017).

Despite advances in supportive care, individuals with SCD and βTM continue to face challenges and reduced life expectancy (Lubeck et al. 2019; Farmakis et al. 2020). While gene therapy trials show promise, hematopoietic stem cell transplantation remains the only established cure, although it is limited to cases with a human leukocyte antigen (HLA)-identical sibling donor (Bhatia and Walters 2008). Pre-implantation genetic testing (PGT) offers an alternative by enabling the selection of unaffected embryos before implantation, thereby avoiding the need for pregnancy termination (Xu et al. 1999). Additionally, PGT plays a crucial role within the hemoglobinopathy community, as it can identify embryos that are HLA-identical to a living child with SCD or βTM, ensuring a matched donor (Basille et al. 2009).

Although PGT has been available for over two decades, its utilization remains limited among parents of children with conditions like SCD. It can be performed for monogenic disorders or single gene defects, with HLA typing applied for various indications, among which hemoglobinopathies like β-thalassemia are

most common (Mamas et al. 2022). However, cultural and religious factors significantly influence decision-making regarding prenatal diagnostic services, making education and counseling crucial (Bruwer et al. 2014). Recent studies indicate a lack of awareness and varying levels of acceptance concerning alternative reproductive options to mitigate the risk of transmitting genetic conditions like SCD and βTM (Attia et al. 2020; Kripalani et al. 2019; Schultz et al. 2020; Alsalman et al. 2023; Al-Kharusi et al. 2021; Bruwer et al. 2022; Darbari et al. 2018; Combs et al. 2023). The importance of education, sensitive to individual preferences and ethical considerations, cannot be overstated. Furthermore, economic factors and healthcare accessibility may significantly influence parental perspectives (Schultz et al. 2020; Combs et al. 2023). This study aimed to investigate knowledge, attitudes, and practices related to PGT among a sample of parents of children with SCD and βTM in Muscat Governorate, Oman. By exploring these factors, we seek to contribute to a more informed and nuanced understanding of reproductive decision-making in this specific context.

METHODS

A questionnaire-based survey was conducted by the principal investigator, researchers, and trained data collectors, including general practitioners and staff nurses from primary healthcare units and the hematology clinic at the Royal Hospital in Muscat Governorate. The Royal Hospital, which houses the National Genetics Center—a leading facility for genetic services in Oman—served as the primary site for data collection. Parents of children with SCD or βTM who were attending the Royal Hospital's hematology clinic or day care unit for blood or exchange transfusions were recruited. Only Omani parents with biological children diagnosed with SCD or βTM were included in the study. Non-Omani or non-Arabic-speaking parents, as well as non-parental guardians of children with SCD or βTM, were excluded, as were those who did not complete the full interview. The sample size of 385 participants was determined based on a 95% confidence interval, a 5% margin of error, and a large population size, given the unknown number of individuals who would benefit from PGT technology.

A two-part questionnaire was developed by the principal investigator. The first section gathered information regarding the participants' demographic and family characteristics, including their age, gender, education level, income, and total number of children. Additionally, it also enquired about the number of children affected by SCD or βTM among their offspring. The second section assessed participants' knowledge, attitude, and practices related to PGT and premarital screening. This section focused on their awareness of PGT, understanding of its benefits, willingness to undergo the procedure, and factors influencing their decision-making regarding reproductive options.

To ensure the validity of the questionnaire, a content validity index was utilized, with seven experts reviewing the questions. Questions scoring below 0.59 were considered to have low content validity and were removed from the questionnaire. Questions with scores between 0.60 and 0.74 were considered to have moderate content validity and were reviewed by the researchers for potential modifications. A pilot study involving 20 participants was then conducted to further validate the questions. Feedback from both the pilot study participants and the data collectors was analyzed, leading to minor corrections to

enhance the clarity and comprehensibility of the final version of the questionnaire. The internal validity of the version of the questionnaire used in the pilot study was not assessed due to differences in questioning that rendered it inapplicable.

The final version of the survey was administered through face-to-face or telephone interviews. Verbal consent was obtained for the participants, and interviews were scheduled at a convenient time. All interviews were conducted in a quiet and private space to ensure confidentiality. Participants were introduced to the purpose of the interview, and their identities were verified for security purposes. Questions were asked in a clear and neutral manner to allow open responses. Interview data were subsequently entered into a Google Form (Google LLC, Mountain View, CA) by the investigators and data collectors, and responses were regularly checked by the primary investigator to ensure accuracy. At the end of the interview, participants were thanked, and contact information was provided for any further questions or concerns.

All statistical analyses were performed using the Statistical Package for the Social Sciences (SPSS) software, version 29.0 (IBM Corp., Armonk, NY). Continuous variables were presented as means, medians, standard deviations, and interquartile ranges, while categorical variables were reported as frequencies and percentages. An independent samples t-test was used to compare means between two groups, while the Mann-Whitney U test was used to compare medians. Associations between two categorical variables were assessed using a Chi-squared test, either Fisher's exact test or likelihood ratio as appropriate. A p value of less than 0.05 was considered statistically significant.

RESULTS

Table 1 presents the demographic characteristics of the 385 participants. There was a slightly higher proportion of males (53.5%) compared to females (46.5%). Educational backgrounds were diverse, with the majority of participants having high school diplomas (53.1%). Income distribution varied, with participants most frequently earning between 301–600 Omani riyals (OMR; 33%) per month, followed by 601–1,000 OMR (26.3%). The mean and median ages of participants were similar, with ages ranging from 22 to 60 years. Total number of children per participant ranged from 1 to 16, while the number of children affected by SCD or βTM varied from 1 to 8. Just over half of the participants (51.9%) reported a family history of hemoglobinopathies, as marked by the presence of relatives with SCD or βTM.

Variable	n (%)
Age (years), mean ± SD	39.06 ± 6.76
Gender	
Male	206 (53.5)
Female	179 (46.5)
Education level	
Illiterate	2 (0.5)
School	66 (17.2)
Diploma	204 (53.1)
College	81 (21.1)
Postgraduate	31 (8.1)
Monthly income (OMR)	
0	31 (9.1)
<300	29 (8.5)
301–600	113 (33.0)
601–1,000	90 (26.3)
>1,000	79 (23.1)
Missing/Not reported	43 (11.2)
Number of children, median (IQR)	3 (3–4)
Number of children with SCD or βTM, median (IQR)	1 (1–2)
Relatives with SCD or βTM	
No	185 (48.1)
Yes	200 (51.9)

SD, standard deviation; OMR, Omani riyals; IQR, interquartile range; SCD, sickle cell disease; βTM, beta-thalassemia major.

Awareness and Knowledge

The majority of participants (96.4%) were aware that SCD and βTM were inherited genetic disorders. However, most (88.3%) were unaware of the possibility of having a child affected by these conditions

prior to their first pregnancy. Only 41.8% were knowledgeable about the option of PGT as a reproductive strategy to prevent conceiving a child with SCD or βTM. Overall, 31.2% of participants reported having received information about PGT from a healthcare worker. Furthermore, 57.7% of participants recognized that the primary advantage of PGT is its ability to prevent the birth of children affected by genetic disorders.

Table 2 presents associations between PGT-related knowledge and various sociodemographic characteristics. There was no significant association between gender and knowledge of PGT ($p = 0.680$). Conversely, there were statistically significant associations with both education level and monthly income, with participants possessing higher education levels and incomes being significantly more likely to exhibit knowledge of PGT ($p < 0.001$ and 0.007, respectively). There was also a significant association with age, with younger participants being significantly more likely to show awareness of PGT as a reproductive option ($p < 0.001$).

ttitudes

Overall, 77.6% of participants believed that their potential offspring might be at risk for hereditary blood disorders, with 72.5% indicating that this awareness influenced their plans to have additional children in the future. After receiving a brief educational summary about PGT from the researchers and data collectors, most participants (76.1%) reported feeling pleased to have learned about PGT. A considerable proportion (86.4%) expressed acceptance of PGT and considered utilizing it in the future. The vast majority (96.6%) agreed on the importance of informing parents about PGT if there was a likelihood of their children being affected by hereditary blood disorders. Most (89.4%) believed that doctors should discuss PGT with carriers of hereditary blood diseases before the birth of their first child. Additionally, an overwhelming majority (96.9%) supported government regulation of PGT. While 85.2% agreed that society would accept PGT, 87.8% were concerned that the technology could impose psychological or financial burdens on parents.

Practices

The majority of participants (92.2%) had not previously undergone PGT. Among those who had, 26% reported not receiving counseling regarding their results, while 43% experienced physical, emotional, and financial stressors. Table 3 shows associations between prior experience of PGT and selected characteristics. Gender ($p = 0.004$), education level ($p = 0.001$), and monthly income ($p = 0.002$) were associated with an increased likelihood of having undergone the procedure. Additionally, participants who were aware of PGT as a reproductive option were more likely to have undergone the procedure ($p < 0.001$). Interestingly, those who reported having previously undergone premarital screening with their partner were more likely to perceive the risk of having children affected by diseases like SCD and βTM prior to their first pregnancy ($p < 0.001$), as shown in Table 4.

Variable	No (n = 355)	Yes (n = 30)	p value
Age (years), mean ± SD	39.25 ± 6.78	36.80 ± 6.09	0.056
Gender			
Male	198 (96.1)	8 (3.9)	0.004*
Female	157 (87.7)	22 (12.3)	
Education level			
Illiterate	2 (100)	0 (0)	0.001*
School	63 (95.5)	3 (4.5)	
Diploma	196 (96.1)	8 (3.9)	
College	69 (85.2)	12 (14.8)	
Postgraduate	24 (77.4)	7 (22.6)	
Monthly income (OMR)			
0	31 (100)	0 (0)	0.002*
<300	29 (100)	0 (0)	
301–600	109 (96.5)	4 (3.5)	
601–1,000	83 (92.2)	7 (7.8)	
>1,000	67 (84.8)	12 (15.2)	
Number of children with SCD or βTM, median (IQR)	1 (1–2)	1 (1–2)	0.313
Relatives with SCD or βTM			
No	167 (90.3)	18 (9.7)	0.187
Yes	188 (94.0)	12 (6.0)	
Knowledge of PGT			
No	223 (99.6)	1 (0.4)	<0.001*
Yes	132 (82.0)	29 (18.0)	

PGT, pre-implantation genetic testing; SD, standard deviation; OMR, Omani riyals; SCD, sickle cell disease; βTM, beta-thalassemia major; IQR, interquartile range.

*Statistically significant at p <0.05.

Table 4: Association between prior awareness of risk of genetic disorders and premarital screening practice among Omani parents of children affected by hemoglobinopathies (N = 385)

Variable	No (n = 318)	Yes (n = 67)	p value
Awareness of risk of having a child with SCD or βTM prior to first pregnancy			
No	292 (85.9)	48 (14.1)	<0.001*
Unsure	1 (100)	0 (0)	
Yes	48 (71.6)	19 (28.4)	

SCD, sickle cell disease; βTM, beta-thalassemia major.

*Statistically significant at p <0.05.

DISCUSSION

Understanding parental knowledge, attitudes, and practices related to PGT in the context of inherited hematological diseases is essential for ensuring that families have access to comprehensive information, support, and resources to make informed decisions about their reproductive options. The findings of this study revealed a notable gap between the high level of awareness of SCD and βTM as inherited genetic disorders and the relatively low knowledge about PGT among Omani parents of children with SCD or βTM. Despite 96.4% of participants being aware of these genetic disorders, only 41.8% were familiar with PGT as a preventive reproductive strategy, indicating a significant gap in the availability or dissemination of information in Oman concerning genetic selection or screening techniques. This disparity suggests a critical need for enhanced educational efforts to bridge the knowledge gap, particularly among individuals at risk of having children with inherited disorders. The lack of awareness about PGT before pregnancy, as reported by 88.3% of participants, underscores the importance of integrating comprehensive genetic counseling into routine healthcare services to inform prospective parents about available options for preventing genetic disorders.

Our study identified significant associations between sociodemographic factors and knowledge of PGT. Participants with higher levels of education and income, as well as younger individuals, demonstrated greater awareness of PGT. These findings align with previous research suggesting that higher educational attainment and socioeconomic status are correlated with better health knowledge (Zajacova and Lawrence 2018; Adler et al. 1994). The results imply that targeted educational initiatives may be more effective if tailored to specific sociodemographic groups. For instance, enhancing educational outreach in lower-income communities and among older individuals could potentially increase PGT awareness and utilization.

The study also found that 86.4% of participants were willing to accept and consider undergoing PGT in the future, suggesting a high level of acceptance of this technology among Omani parents with children affected by SCD or βTM. This awareness rate is higher than that reported in other countries, such as the

United States and Saudi Arabia, where only 24–44% and 38.5% of parents, respectively, were aware of PGT and only 36.4% of Saudi Arabian participants reported a willingness to undergo the procedure (Attia et al. 2020; Kripalani et al. 2019; Alsalman et al. 2023; Traeger-Synodinos and Harteveld 2017). The majority of participants in the present study (57.7%) perceived the main advantage of PGT as preventing the birth of children affected by inherited disease, which aligns with the primary purpose of PGT, namely to identify and select embryos without genetic disorders.

Healthcare workers play a crucial role in educating individuals about reproductive options like PGT. The fact that 31.2% of participants received information about PGT from a healthcare professional underscores the necessity for healthcare professionals to provide accurate and up-to-date information about genetic testing and reproductive options. This is particularly important for individuals at risk of passing on hereditary blood disorders like SCD and βTM to their future offspring. Additionally, increasing public awareness through various channels such as social media, television, radio, magazines, and websites is vital. Previous studies have highlighted that educational materials and awareness campaigns can positively influence attitudes and practices regarding reproductive options and ensure accurate knowledge (Attia et al. 2020; Kripalani et al. 2019; Schultz et al. 2020).

The study revealed that the vast majority of participants (92.2%) had not previously undergone PGT. Among those who had, over one-quarter (26%) reported not receiving counseling sessions to discuss their results. Moreover, interviews indicated that some participants had received inaccurate information regarding their results. Enhancing pre- and post-test counseling, as well as providing resources to manage the emotional and financial impacts of PGT, could improve the overall experience and encourage more individuals to consider PGT. Participants who had undergone premarital screening were more likely to perceive the risk of having children affected by SCD and βTM ($p < 0.001$), suggesting that awareness of screening technologies may influence perceptions of genetic disease risk in future children. Additionally, participants who were knowledgeable about PGT were significantly more likely to have undergone the procedure themselves ($p < 0.001$), indicating that awareness of PGT may impact uptake of this reproductive option.

The study found that 43% of participants who underwent PGT experienced physical, emotional, and financial stressors, while 36.6% reported not facing any stressors. Other studies in Oman have similarly noted that the PGT process can be physically exhausting and emotionally draining (Al-Kharusi et al. 2021; Bruwer et al. 2022). Among those who did not accept PGT, 33.3% cited financial reasons, 25% cited religious reasons, and 25% cited personal reasons. Acceptance of PGT is influenced by multiple factors, including socioeconomic status, religion, epidemiological determinants, and the prevalence of autosomal recessive conditions (Traeger-Synodinos and Harteveld 2017; Malik et al. 2022). Addressing these concerns through supportive counseling and financial assistance programs could mitigate barriers to PGT adoption and ensure that individuals can make informed and accessible reproductive choices.

The study highlights the need for healthcare providers to proactively incorporate discussions about PGT into routine genetic counseling and prenatal care. Policy recommendations include advocating for

increased funding for public education campaigns about genetic disorders and PGT. Additionally, integrating PGT discussions into routine prenatal screening protocols could normalize and increase uptake of these technologies.

The findings of this study should be interpreted with consideration of several limitations. The cross-sectional design provides a snapshot of data at a specific point in time, which limits the ability to draw causal conclusions or track changes over time. Cultural, social, and religious barriers may have impacted the accuracy of responses. Additionally, the study concentrated on specific aspects of parental reproductive decision-making related to SCD and βTM, potentially overlooking other relevant factors, such as the cost comparison between screening for hematological and other genetic disorders versus in vitro fertilization and PGT in Oman. Future research should investigate knowledge, attitudes, and practices related to PGT during the premarital screening stage and among individuals affected by other genetic disorders to achieve more generalizable results. Potential interventions to enhance parental knowledge, attitudes, and practices regarding PGT could include awareness campaigns targeting healthcare workers and the general population, increased media coverage, and the implementation of premarital screening in primary healthcare centers. Moreover, the findings of this study call for increased integration of genetic counseling into routine healthcare services.

CONCLUSIONS

The study revealed a lack of knowledge of PGT among Omani parents of children with SCD and βTM, emphasizing the urgent need for enhanced education and awareness about PGT and other reproductive options for individuals at risk of genetic disorders. Despite this, a high level of acceptance of PGT was observed among these parents, highlighting the potential benefits of this technology in preventing hereditary blood disorders. However, the study also identified various barriers to PGT acceptance, including physical, emotional, and financial stressors, as well as religious and personal reasons. These findings underscore the importance of not only raising awareness about PGT and related reproductive options but also addressing the psychological and financial challenges associated with PGT technology. This approach is crucial to ensuring that PGT is used effectively and ethically in reproductive decision-making.

Declarations

Competing Interests

The authors declare no competing interests. The authors have no financial or non-financial interests that are directly or indirectly related to the work submitted for publication.

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Ethical Approval

This study received ethical approval from the Ministry of Health Center for Research and Studies (Reference: MOH/CSR/23/27628). All procedures adhered to the Declaration of Helsinki and Omani national guidelines for human subjects research.

Data Availability

The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

Informed Consent

Verbal informed consent was obtained from all participants prior to participation in this study. Participants were informed of the study's purpose, procedures, risks, and benefits. Participation was entirely voluntary, and participants could withdraw at any time without penalty.

Author Contribution

Author ContributionsAsma Al-Musalhi (AAM): Conceived and designed the study, developed the questionnaire, oversaw data collection, performed statistical analysis, and drafted the manuscript.Salwa Al Obaidani (SAO): Provided clinical genetics expertise, contributed to questionnaire validation and design, and reviewed the result reviewed the manuscript .Alya Al-Hsani (AAH): contributed to questionnaire validation and design, and reviewed the result, reviewed the manuscript Nadhira Al-Batashi (NAB): Conducted face-to-face and telephone interviews, collected data reviewed the manuscript .Sara Al-Busafı (SAB): Conducted face-to-face and telephone interviews, collected data, reviewed the manuscript.Manar Al-Maawali (MAM): Conducted face-to-face and telephone interviews, collected data , reviewed the manuscript.Zainab Al-Hinai (ZAH): Conducted face-to-face and telephone interviews, collected data, reviewed the manuscript.Suad Al-Habsi (SAH): Coordinated nursing staff and participant recruitment at the hematology clinic and day-care unit, reviewed the manuscript.Zwaina Al-Qasabi (ZAQ): Conducted face-to-face and telephone interviews, collected data, reviewed the manuscript.Ashjan Al Mamry (AAM): Conducted face-to-face and telephone interviews, collected data, reviewed the manuscript.

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