





## SPECIAL ARTICLE

# Qatar's genetic counseling landscape: Current insights and future prospects



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### ABSTRACT

Genetic counseling is a dynamic and rapidly growing field. In Qatar, the significance of genetic counseling is underscored by the distinctive demographic characteristics of the population, including elevated rates of consanguinity and larger family sizes, contributing to the increased incidence of many genetic conditions. This emphasizes the crucial role of genetic counseling in addressing the specific needs of the community. Over the past decade, key health care institutions in Qatar, such as Hamad Medical Corporation and Sidra Medicine, have significantly expanded genetic counseling services encompassing premarital, reproductive, prenatal, pediatric, adult, and cancer care. This multifaceted approach reflects Qatar's health care system's commitment to addressing various aspects of genetic health and well-being across different life stages.

A pivotal milestone in the field's development in Qatar was the establishment of a genetic counseling master's program at Qatar University in 2018, showcasing the country's dedication to fostering indigenous expertise in genetic counseling with the necessary competencies and cultural sensitivity to address the unique genetic counseling needs of the population. The recognition of genetic counseling as a profession and the licensure by the Ministry of Public Health in Qatar is another key achievement to ensure the high quality of service and protection of the profession. Contributing to global genetic knowledge, various academic and research entities in Qatar are conducting genetic/genomic/genetic counseling research toward advancing precision medicine in the country, and initiatives such as the Qatar Biobank and Qatar Genome Program have played a major role in catalyzing these efforts.

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## Introduction

Genetic counseling is a dynamic and evolving field that plays a crucial role in the interaction of genetics and health care. As outlined by the National Society of Genetic Counselors, “Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease.”<sup>1</sup> Genetic counselors primarily engage in clinical roles, as defined by the National Society of Genetic Counselors. However, an expanding number of genetic counselors are currently involved in diverse positions and settings, such as research, laboratories, education, and advocacy across different regions.<sup>1</sup>

In 1947, the term “genetic counseling” was first introduced by Sheldon Reed, who considered it as a form of social work meant to benefit individual families rather than prioritizing the interests of the state or the population. From the 1940s to 1960s, early practitioners primarily offered genetic advice, and many of these early practitioners were nonclinical scientists.<sup>2</sup> The history of genetic counseling education started in 1969 in New York with the first genetic counseling master’s program that was established at Sarah Lawrence College.<sup>3</sup> Since that time, the profession has been expanding around the globe. The role of genetic counselors became even more critical with the mapping of the human genome in the Human Genome Project, which was completed in 2003. This monumental achievement provided a comprehensive understanding of the DNA sequences that make up the human genome. Genetic counselors became essential for bridging the gap between the scientific community and the general population, helping individuals comprehend the implications of genomic information on their health and the health of their offspring.<sup>4</sup>

Genetic counseling encompasses a wide range of services tailored to meet the unique needs of individuals and families. These services include risk assessment for genetic conditions, education about genetic testing options, interpretation of test results, guidance on reproductive choices, family planning, and providing psychosocial support. Genetic counselors are now part of multidisciplinary teams, contributing to the new personalized medicine approach in health care.<sup>5</sup> The expansion of genetic testing technologies has significantly influenced the landscape of genetic counseling. From chromosomal analysis to single-gene testing and through to comprehensive and advanced DNA sequencing, such as next-generation sequencing (NGS), there has been significant progress. This advancement in NGS has enabled more precise identification of genetic variations associated with various conditions. Consequently, genetic counseling now extends its scope to encompass not only individuals with established genetic conditions but also those at risk, considering factors such as family history.<sup>5</sup>

In looking at genetic counseling from a local perspective in Qatar, it is crucial to highlight the distinct features and the population dynamics within the country. Qatar, situated on

the Arabian Peninsula, showcases unique demographics, cultural intricacies, and a health care system that influences the field of genetic counseling. Examining the details of genetic counseling in this dynamic Gulf state provides valuable insights into the specific challenges and opportunities shaped by the local cultural and health care context.

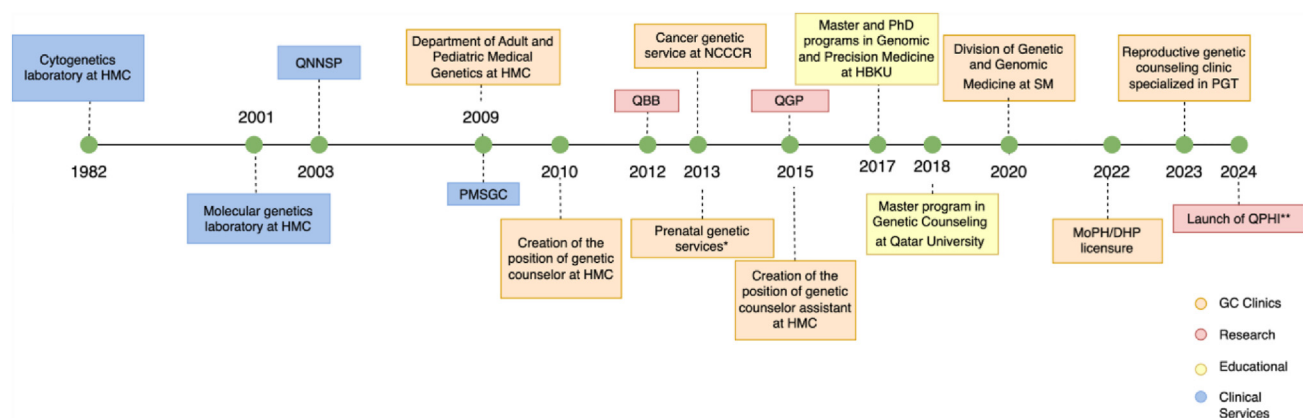
This article aims to provide a comprehensive exploration of the journey of genetic counseling in Qatar. We explore here the historical roots, the evolution, and the unique aspects defining genetic counseling and offer insights into how genetic counseling services have been tailored to meet the specific needs of individuals and families in Qatar.

## Demographics of Qatar

Qatar is located on the Arabian Peninsula, an extension that stretches northward from the eastern edge of the Arabian Peninsula into the Arabian Gulf, covering an area of 11,427 square kilometers. The population of Qatar is approximately 2,966,000 million people,<sup>6</sup> which includes 600,000 Qataris. The population of Qatar exhibits a notable prevalence of consanguineous marriages. A previous study found that over 54% of marriages among native Qataris are consanguineous, with first-cousin marriages being the most common at 34.8%.<sup>7</sup> Another study conducted in Qatar on 599 Qatari families with genetic and nongenetic conditions recruited from 2 centers, Hamad Medical Corporation (HMC) and the Shafallah Center for Children with Special Needs, revealed that consanguineous marriages were prevalent in 66.2% of the families, underscoring the significant impact of consanguinity on such conditions.<sup>8</sup> Consanguinity is also prominent in various expatriate communities residing in Qatar. In conjunction with the inclination toward larger family sizes and an extended reproductive period, consanguinity contributes to the increased prevalence of several autosomal recessive conditions within the Qatari population, such as homocystinuria, cystic fibrosis, and hemoglobinopathies.<sup>9</sup>

## Historical perspective on the evolution of genetic counseling in Qatar

Genetic counseling in Qatar has undergone a remarkable evolution, shaped by a confluence of cultural, scientific, and health care advancements. This historical journey is marked by key milestones, developments, and the contributions of influential figures and organizations, all of which have played significant roles in shaping the landscape of genetic counseling in this dynamic and culturally rich Gulf state (Figure 1). The establishment of genetic counseling services in Qatar reflects a proactive approach to addressing the genetic aspects of health and disease within the population especially because of the increased demands of the population due to the special demographic features discussed above. As the field gained recognition globally, Qatar recognized the importance of incorporating genetic



**Figure 1** A timeline for the major advancements in the field of genetics and genetic counseling in Qatar. \*Previously provided at the Women's Hospital at HMC until 2017 and then transferred to WWRC in 2018 at HMC. \*\*The formal establishment of QPHI was in 2020.

counseling services into its health care system to empower individuals and families with the knowledge needed to make informed decisions about their genetic health.<sup>9</sup>

The genetic counseling landscape in Qatar has experienced noteworthy advancements led by health care leaders, HMC and Sidra Medicine (SM). HMC is a governmental institution that serves as Qatar's primary provider of advanced health care services and stands as a prominent network of hospitals in the Middle East. HMC was at the forefront of integrating genetic counseling services into the health care framework.<sup>9</sup> Genetic clinics were first established in 2002 and evolved into a well-structured department for genetic services in 2009, which was named the Department of Adult and Pediatric Medical Genetics and paved the way for the introduction of genetic counseling clinics in the country in 2011. These clinics provided comprehensive genetic counseling services and were made accessible to patients with a broad range of conditions and indications.<sup>9</sup> HMC remained the sole genetic counseling service provider for all specialties until 2020.

SM is another institution dedicated to delivering comprehensive health care services for children and women in Qatar. A notable milestone took place in 2020 when pediatric genetic counseling services were transferred to SM alongside the establishment of the Division of Genetic and Genomic Medicine at SM.

The clinical genetics teams from both HMC and SM across specialties currently collectively consist of 10 master's trained genetic counselors, 4 genetic counseling assistants, 4 clinical and metabolic geneticists, and 4 clinical and metabolic genetic fellows who work collaboratively to provide a spectrum of genetic counseling services for the whole country. This unified approach solidifies their position as key providers of comprehensive genetic care, spanning premarital, reproductive, prenatal, pediatric, adult, and cancer genetic services, ensuring a holistic and integrated genetic health care framework in Qatar.

Qatar's genetic counselors constitute a diverse group, including 4 Qatari and 6 non-Qatari (Arab) genetic counselors, each possessing unique qualifications and areas of

expertise as described in Table 1. Collectively, the team offers a wealth of knowledge and expertise to deliver comprehensive genetic counseling services across various specialties in Qatar. It is worth noting that genetic counselors who are currently providing pediatric and adult genetic counseling services at SM and HMC, respectively, cover a wide range of subspecialties, including cardiology, nephrology, immunology, dermatology, pulmonology, endocrinology, rheumatology, audiology, ophthalmology, and neurology. Moreover, all team members currently operate within clinical settings, with plans to integrate genetic counselors into laboratory settings, in the near future, to enhance the scope of the profession and the efficiency of services provided.

Moreover, the availability of advanced genetic testing technologies stands out as a crucial development globally. Within Qatar, the Diagnostic Genomic Division (DGD) at the Department of Laboratory Medicine and Pathology in HMC is an accredited lab by the College of American Pathologists, and it provides the majority of genetic testing services. The DGD lab includes the cytogenetic and the molecular genetics laboratory sections. Furthermore, in the past 10 years, cancer services have been established within DGD to test patients with solid tumors and hematological malignancies. The DGD provides genetic services, handling referrals from all over Qatar for diagnostic purposes related to hereditary conditions prevalent in the population. The facility conducts tests in areas such as prenatal diagnostics (eg, for beta-thalassemia and sickle cell disease), postnatal (eg, for fragile X syndrome and Y-chromosome microdeletions), and premarital (eg, cystic fibrosis and spinal muscular atrophy [SMA]). The available methodologies within DGD include karyotyping, fluorescent in situ hybridization, chromosomal microarray analysis, polymerase chain reaction, real-time PCR, droplet digital PCR (ddPCR), DNA methylation, fragment analysis, DNA sequencing by Sanger sequencing, and NGS for targeted gene panels. The exome sequencing (ES) is a valuable tool, especially for unexplained cases in which a Mendelian disorder is suspected. Moreover, the DGD lab provides molecular testing

**Table 1** Characteristics of all genetic counselors in Qatar ( $N = 18$ )

Category	Number
Qualification	
International master's program	7
- USA	4
- UK	2
- Canada	1
Local master's program	11
- Qatar	11
Total	18
Licensure	
Licensed by MoPH	8
Eligible for MoPH License	10
Total	18
Credential	
International credential	6
- CGC by ABGC	4
- CCGC by CBGC	2
No international credential	12
Total	18
Area of practice	
Clinical <sup>a</sup>	11 <sup>a</sup>
- Adult	4
- Reproductive	4
- Cancer	5
- Pediatric	3
- Prenatal	2
- Other (nursing)	1
Academic	1
- Genetic counseling master program	1
Research	2
- Qatar University	1
- HMC <sup>a</sup>	1 <sup>a</sup>
- QPHI (QGP <sup>b</sup> )	1
In different field	4
Total	18

ABGC, American Board of Genetic Counseling; CBGC, Canadian Board of Genetic Counseling; CCGC, Canadian Certified Genetic Counselor; CGC, Certified Genetic Counselor; HMC, Hamad Medical Corporation; MoPH, Ministry of Public Health; QGP, Qatar Genome Program; QPHI, Qatar Precision Health Institute; UK, United Kingdom; USA, United States of America.

<sup>a</sup>Several Genetic Counselors are practicing in different areas.

<sup>b</sup>QPHI includes QGP.

as part of 3 national screening programs, (1) National Pre-marital Screening and Genetic Counseling Program (PMSGC), (2) Qatar National Newborn Screening Program, and (3) breast cancer screening program. All positive cases from these 3 screening programs are referred to the relevant genetic counseling clinic, for instance, newborn cases are referred to the pediatric clinics at SM, whereas premarital cases are referred to the adult clinics at HMC. Therefore, these programs resulted in increasing the demand for genetic counseling services and genetic counselors to achieve the intended outcomes from these national programs.

Moreover, the DGD lab outsources some genetic tests to international laboratories. This strategic collaboration ensures that Qatar has access to a broader spectrum of genetic testing

methodologies and expertise, particularly for rare or complex genetic conditions that may require specialized equipment or specific technological platforms available in those overseas laboratories.<sup>9</sup> Simultaneously, SM's Pathology Genetics laboratory contributes to Qatar's genetic testing supply. With in-house capabilities for ES and genome sequencing (GS), SM's lab enhances the repertoire of genetic testing services in the country. Similar to the DGD lab, SM's Pathology Genetics also partners with external laboratories overseas to facilitate genetic tests not performed in house.

Genetic counseling services in Qatar are primarily government provided and accessible to all citizens and residents for a minimal charge or no charge. Additionally, genetic testing is fully covered by the government for all citizens. As for residents, genetic tests that are well established within the country, such as karyotype and chromosomal microarray analysis, are offered at no cost, whereas more advanced genetic tests that are locally labor intensive or that need to be performed at abroad laboratories, such as multi-gene panels, ES, and GS require out-of-pocket payment or alternative methods of financing, such as charity support.

Another significant milestone in the genetic counseling journey in Qatar was the establishment of formal education and a graduate master's training program in genetic counseling at Qatar University (QU). This demonstrates Qatar's dedication to fostering local expertise in the domain, guaranteeing a reservoir of capable and culturally aware genetic counselors to fulfill the increasing need for such services.

Further contributing to Qatar's genetic landscape are notable research initiatives. The Qatar Biobank (QBB) and Qatar Genome Project (QGP) both are currently part of Qatar Precision Health Institute (QPHI), are pioneering endeavors, advancing our understanding of the genetic underpinnings of various health conditions within the Qatari population toward the implementation of precision medicine in the country. Importantly, QGP has a dedicated genetic counselor in house, playing a crucial role in providing genetic counseling services to QGP participants. QBB and QGP have generated GS data for more than 30,000 Qataris with comprehensive phenotypic data that facilitate the understanding of the genetic map of the Qatari population and the genetic variants underlying the various genetic conditions in Qatar.<sup>10-13</sup> Another QGP/QBB initiative aimed to enhance the molecular diagnosis capacity for numerous diseases through the development of QChipPMv2, an SNP array designed for detecting over 50,000 known pathogenic variants associated with a broad spectrum of genetic disorders, including variants identified in Qatar. This innovative tool aims to provide accurate and comprehensive screening for such an understudied population and facilitate the implementation of precision medicine.<sup>14</sup>

## Genetic counseling services and programs in Qatar

In the rapidly advancing field of genetic counseling in Qatar, genetic counselors and medical geneticists play a pivotal

role across various life stages, addressing a spectrum of genetic concerns and conditions.

In Qatar, the clinical genetics team works seamlessly alongside multidisciplinary teams, combining clinical expertise in medical, psychosocial, and case management domains. Although their foundational education equips them as generalists, genetic counselors and medical geneticists have the flexibility to specialize in areas such as premarital, reproductive, prenatal, pediatric, adult, and cancer services, and they are currently covering more than 1 area of practice. Such specialization allows them to provide tailored guidance specific to the intricacies of genetic counseling services.

### **Prenatal genetic counseling**

Prenatal genetic counseling services form an integral component of the prenatal care currently provided at the Women's Wellness Hospital and Research Center (WWRC), which is part of HMC. Prenatal genetic services were established back in 2013 as a part of the genetic services and are targeting women with high-risk pregnancies, history of abnormal pregnancies, or a family history of genetic conditions.<sup>9</sup> The genetics team plays a significant role in the collaborative efforts within the Fetomaternal Medicine Unit (FMU) at WWRC. This unit is responsible for managing pregnant females with high-risk pregnancies due to obstetric, medical, surgical, or genetic complications, and they conduct about 7000 examinations for women referred all across the country yearly.<sup>15</sup> The Prenatal Genetics Clinic operates once per week and is managed by 1 medical geneticist and 1 or 2 genetic counselors who provide comprehensive genetic counseling services to high-risk pregnant women. The primary objective of the prenatal counseling sessions is to discuss screening and diagnostic tests and their outcomes, provide crucial insights into potential genetic conditions, and support informed decision making regarding current or future pregnancies. During the genetic counseling sessions, a geneticist and genetic counselors collaborate to provide comprehensive guidance to referred pregnant women regarding their current pregnancies. This is facilitated by the FMU obstetricians and gynecologists through the utilization of prenatal screening methods, such as ultrasound (US) and prenatal cell-free DNA screening.<sup>16</sup> All pregnant women are offered routine screening by US as part of standard practice. Prenatal screening blood tests, including prenatal cell-free DNA screening, are offered by obstetricians in the governmental system for particular clinical indications, such as advanced maternal age and abnormal US screening. In the private sector, obstetricians may offer prenatal screening blood tests to all pregnant women. The prenatal genetics clinic provides a diagnostic service whereby only diagnostic prenatal genetic testing is offered to and facilitated for pregnant women with a variety of indications.<sup>16</sup> Prenatal diagnostic tests encompass a range of methodologies aimed at identifying potential issues during pregnancy, such as karyotyping and/or chromosomal microarray analysis. Furthermore, more

advanced diagnostic testing options are available, such as gene panels and ES, offering a more comprehensive analysis of genetic information to uncover potential genetic conditions or variations during the prenatal stage.<sup>16</sup> By working closely with the FMU team, this collaborative approach ensures a seamless integration of genetic counseling services into the broader spectrum of comprehensive care provided to pregnant women.

### **Pediatric genetic counseling**

Pediatric genetic counseling services are currently provided at the Division of Genetic and Genomic Medicine at SM. Genetic services and counseling, facilitated by a team of 3 genetic counselors, 4 clinical and metabolic geneticists, and 4 clinical and metabolic genetics fellows, are tailored to address the unique needs of pediatric patients under the age of 18 years who present with diagnosed or suspected genetic conditions or with a family history of genetic conditions in addition to cases with birth defects, dysmorphic features, and/or developmental delay. Pediatric genetic counselors at SM receive referrals from all medial subspecialties across SM and HMC, as well as external referrals. There are special pediatric care aspects in Qatar because of its unique demographical factors, such as large family size and high consanguinity rates that favor the incidence of autosomal recessive conditions, contributing to complex scenarios with the presence of 2 or more genetic conditions in the same family, in the same sibship, or even in the same patient. This specialized pediatric genetic care encompasses a multifaceted approach, emphasizing extensive counseling and education for parents aiming to resolve such complex cases.

Patients are presented with the different possible options for diagnostic genetic testing to confirm or rule out suspected clinical diagnoses and the selection of genetic tests is tailored to each case. In situations which there is a documented family history of a particular condition, the recommendation may be to undergo targeted testing for a known familial pathogenic variant in a single gene. Alternatively, in cases without relevant family history, genetic testing options may include more comprehensive tests, such as gene panels or ES. The customization of genetic testing based on the patient's unique circumstances underscores a personalized approach to diagnostic evaluations. The focus lies not only on explaining the details of the genetic diagnosis affecting the child but also on guiding parents through the management plan. This involves specifying the appropriate medical specialties to follow with, outlining the care required, and presenting options for genetic diagnostic testing. In addition to comprehensive counseling, parents are provided with crucial information regarding the recurrence risk of genetic conditions in future children and the identification of other at-risk family members. The genetic counselors also discuss reproductive options, including preimplantation genetic testing (PGT) and prenatal diagnosis. Moreover, the genetics team collaborates with research entities such as SM clinical research to attempt to diagnose undiagnosed/unsolved cases following a translational research approach.

### Adult genetic counseling

Genetic services and counseling for adult patients in Qatar are provided at the Department of Adult and Pediatric Medical Genetics at HMC, facilitated by a team of 4 genetic counselors in addition to the 4 clinic and metabolic geneticists within the team. Adult genetic services are tailored to address the unique needs of adult patients referred from various medical subspecialties, such as neurology, endocrinology, cardiology, and nephrology. The genetic team works closely with individuals, tailoring the genetic counseling process to the specific medical context of each case. The diagnostic journey involves targeted testing, gene panel testing, or ES and genetic counselors interpret and explain the test results, providing insights into the genetic factors influencing patients' health. Such discussions extend to potential management plans, including referrals for appropriate medical specialties. Beyond case diagnosis and management, genetic counselors engage in comprehensive discussions regarding the recurrence risk of genetic conditions in future family members. These conversations cover various reproductive options, with a particular focus on PGT, especially relevant for individuals considering future pregnancies.<sup>17</sup> Furthermore, a specialized reproductive genetic counseling clinic, emphasizing PGT, was established in 2023 at WWRC within the Section of Reproductive Medicine at HMC.

The adult genetic counseling service also addresses reproductive considerations for adult patients. Healthy ES is a currently available test for married couples who seek insights into their carrier status and shared reproductive risks. Unlike the PMSGC, which is mandatory for couples intending to marry and primarily focuses on identifying the positive carrier status of genetic conditions before marriage. Healthy ES is offered optionally for couples who are already married and concerned about their carrier status because of a known/unknown hereditary condition in the family and/or increased anxiety about the risk of having an affected child with a hereditary condition. Healthy ES involves the use of ES technology to analyze the entire coding region of a person's genome with a focus on autosomal recessive and X-linked conditions. By integrating these various genetic counseling services, Qatar demonstrates a commitment to fostering a healthier, more informed society in which individuals and couples are equipped to make informed decisions about their health and family planning.

### National screening programs

Qatar was one of the first Arab nations to establish comprehensive genetic screening programs, showcasing its commitment to proactive health care initiatives.<sup>9</sup> One notable achievement is the Qatar National Newborn Screening Program, initiated in 2003 through a collaborative partnership with the University Children's Hospital of Heidelberg, Germany.<sup>9</sup> This program encompasses screening all newborns in Qatar during the first 36 to 72 hours of their life for 34 core conditions, such as hemoglobinopathies, metabolic diseases, and other genetic

diseases, for the purpose of early detection and effective management of potentially debilitating health issues in newborns. The advent of tandem mass spectrometry in newborn screening (NBS) has revolutionized global screening programs, allowing for the simultaneous detection of over 20 monogenic disorders in a single diagnostic run. Cases with positive findings are subsequently confirmed through genetic testing.<sup>18</sup> Beyond the evident health benefits, such initiatives also contribute to alleviating the financial strain on the health care system by addressing conditions proactively.<sup>9</sup>

In tandem with the NBS, Qatar implemented a mandatory PMSGC in 2009, which includes both genetic testing and sexually transmitted diseases testing.<sup>19</sup> The genetic testing aims to identify a positive carrier status for highly prevalent genetic conditions in Qatar, including hemoglobinopathies, cystic fibrosis, classical homocystinuria, and SMA. Although the core screenings for the specified disorders are mandatory, SMA screening is offered as an optional test within the program.

Premarital genetic testing in Qatar is mandatory for all Qatari couples intending to marry in the country because it mainly tests for founder variants in the population.<sup>20</sup> It is a prerequisite to be tested to obtain the marriage certificate/approval. In cases which partners identified to both carry the same genetic condition, they still have the option to proceed with the marriage, emphasizing the voluntary nature of this decision irrespective of the test results. Premarital testing is currently provided by 8 primary health care centers across Qatar. This program was initiated to identify carriers of the most common founder genetic variants in the Qatari population: p.I1234V in *CFTR*, p.R336C in *CBS*, and copy-number variants of *SMN1* and *SMN2* genes.<sup>9</sup> Samples collected during this process are exclusively analyzed in HMC's molecular genetics laboratory. Individuals identified with positive findings, indicating that they have a positive carrier status for a specific condition, are subsequently referred to the Department of Adult and Pediatric Medical Genetics at HMC for further genetic counseling.<sup>21</sup> Genetic counselors play a crucial role in premarital genetic counseling, addressing concerns related to education about genetic conditions, consanguinity, and raising awareness about potential hereditary risks. Furthermore, genetic counselors engage in discussions with couples about the latest reproductive options, such as PGT, as a proactive preventive measure, in case they wish to continue with the marriage.<sup>21</sup>

### Cancer genetic counseling

Cancer genetic counseling services in Qatar are predominantly delivered at the National Center for Cancer Care and Research within HMC in hereditary cancer and high-risk clinics, where a comprehensive approach is undertaken in line with the guidelines set forth by the National Comprehensive Cancer Network.<sup>22</sup> In 2013, the clinic initially focused on managing individuals with hereditary breast and ovarian cancers, and the cancer genetic team consisted of 5 genetic counselors. However, as of 2016, the clinics

underwent an expansion to include the management of patients at elevated risk for gastrointestinal and various hereditary cancer syndromes. Referrals to high-risk clinics commonly originate from different departments at HMC, primary health centers, and private hospitals. Patients are typically referred based on a personal history of early-onset cancers such as breast, ovarian, and colon cancers, extensive family history of diverse malignancies, or exposure to specific risk factors.<sup>23</sup> In the cancer clinic, patients undergo various genetic tests tailored to their specific cases. Diagnostic, predictive, and confirmation of the genetic alteration from the high-risk clinic is exclusively done by the genetic counselor in the clinic. Referrals to the molecular genetic laboratory in HMC will reject testing any sample sent directly from any other clinic due to lack of genetic counseling involvement. Those with documented familial pathogenic variants typically undergo targeted familial testing. Alternatively, patients without specific familial variants are commonly recommended gene panel testing or ES.

For affected cancer patients, genetic testing plays a pivotal role in the overall management of their medical journey, beyond understanding the genetic cause. For example, by knowing the genetic cause, oncologists can collect valuable information about specific genetic variants and their various cancer-associated features. This information is instrumental in tailoring a personalized medical plan that aligns with the unique genetic characteristics of the patient's cancer.<sup>22</sup> For instance, utilizing diagnostic *BRCA* analysis is a potent method to identify the subpopulation that is most likely to benefit from personalized therapeutic intervention. In cancer patients with variants in the *BRCA* gene, poly-ADP-ribose polymerase inhibitors have shown notable effectiveness. These inhibitors exploit specific molecular vulnerabilities, providing a precise and promising therapeutic strategy for such cases.<sup>24</sup>

Furthermore, individuals are offered the opportunity to undergo predictive testing, allowing for the identification of potential genetic predispositions, such as hereditary breast and ovarian cancer syndromes related to *BRCA* variants, as well as Lynch syndrome. Early detection and intervention for these conditions contribute to their clinical manageability and positively influence public health outcomes.<sup>25</sup> This comprehensive genetic counseling framework does not only address the immediate needs of cancer patients but also extends its reach to encompass risk-reducing strategies and early detection/prevention, such as risk-reducing surgeries and chemoprevention for those at risk. As an additional preventive measure, these individuals can opt to participate in annual screening programs. The screening programs are designed to monitor their health closely, promptly address any emerging symptoms, and implement proactive measures in case of potential concerns,<sup>23</sup> in addition to offering high-risk families reproductive options, such as PGT and prenatal testing. In Qatar, when parents are positive for genetic variants linked to adult-onset conditions, including cancer syndromes, PGT is an available option that can be combined with assisted reproductive technologies to

screen embryos for heterozygosity or homozygosity for disease-causing variants, allowing informed decisions about which embryos to transfer. Prospective parents who are both positive for a cancer syndrome and have a risk of having a child with a homozygous early-onset condition also have the option of prenatal diagnosis to rule out homozygous status in an ongoing pregnancy. Heterozygosity for adult-onset cancer syndromes is not assessed nor reported as part of prenatal diagnosis to protect the child's future autonomy, deferring nonhomozygous testing decisions until adulthood. Qatar's provision of PGT and prenatal testing serves as a valuable tool for family planning, ensuring that individuals have access to comprehensive genetic screening and reproductive options. By aligning with established guidelines and adopting a patient-centric approach, the genetic counseling services at National Center for Cancer Care and Research contribute significantly to the overall management and care of individuals affected by cancer and those at risk within the familial context.

## Education and Training

In 2018, recognizing the escalating demand for genetic counseling services in Qatar, the College of Health Sciences at QU took a pioneering step by establishing the country's first genetic counseling program. The program was created to address the need for competent and culturally sensitive genetic counselors within the Qatari health care landscape, as well as in the whole region. This is the first genetic counseling master program in Qatar, and it is the third in the Arab world. The previous 2 master's programs were both in Saudi Arabia at Al-Faisal University and King Abdulaziz University, making Qatar the second country in the Arab world and the Middle East and North Africa region.<sup>26</sup>

QU's 2-year master's program is comprehensive, consisting of 46 credit hours (Table 2). It aims to provide students with a well-rounded education encompassing both theoretical and practical courses, equipping them with the necessary competencies for a successful career in genetic counseling. The program focuses on developing a strong foundation in understanding, interpreting, analyzing, and practicing various aspects and subspecialties within the field of genetic counseling. The program plays a pivotal role, being strategically structured to provide students with evidence-based practice skills covering fundamental and advanced genetic counseling principles and techniques, molecular and medical genetics, genomics, ethics, and research skills.

Simultaneously, students are immersed in a dynamic learning environment where they engage in 3 clinical practice courses conducted at HMC and SM. These clinical rotations are designed to offer students real-world genetic counseling experiences in different specialties under the supervision of licensed genetic counselors and medical geneticists. These rotations, extending across various genetic counseling services in Qatar, encompass premarital,

**Table 2** Study plan for the masters in genetic counseling

Term	Course #	Course Title	Credit Hours
First Year (25 Credit Hours)			
Fall	GENC621	Genetic Counseling Practicum I	3
	GENC610	Principles of Genetic Counseling	3
	GENC611	Cytogenetics Developmental Biology	2
	GENC612	Medical and Human Genetics I	3
Total			11
Spring	BIOM515	Molecular Diagnostic	3
	GENC622	Genetic Counseling Practicum II	2
	BIOM540	Research Methods in Biomedical Sciences	3
	GENC613	Medical and Human Genetics II	3
Total			11
Summer Semester			
Fall	GENC640	Clinical Practice I	3
Second Year (21 Credit Hours)			
Fall	GENC620	Advanced Principles of Genetic Counseling	3
	BIOM550	Medical Laboratory Laws and Ethics	3
	GENC650	Clinical Practice II	4
Total			10
Spring	GENC615	Seminar in Genetic Counseling	1
	GENC660	Clinical Practice III	4
	GENC623	Research Project	3
	ELECTIVE	Elective Course <sup>a</sup>	3
	GENC625	Professional Development Exam	0
Total			11

<sup>a</sup>Current elective courses include Advanced Special Topics in Genetic Counseling, Social and Behavioral Science, Health Services Management and Leadership, Current Issues in Clinical Laboratory Sciences, and Health Informatics.

reproductive, prenatal, pediatric, adult, metabolic, and cancer, in addition to a rotation at the DGD lab. Within these clinical environments, students actively partake in providing genetic counseling to patients, facilitating educational initiatives, and delivering essential psychosocial support. Students also navigate the intricate landscape of patient interactions, ensuring a holistic and compassionate approach to genetic counseling services. It is noteworthy that all members of the genetic teams in both SM and HMC are actively contributing to the clinical rotation and training of students, including the graduates from the QU master program who have gained sufficient experience to be involved in training students. Additionally, 1 genetic counselor holding the Canadian Board of Genetic Counseling works as a lecturer at QU and is dedicated full time to the MSc program, focusing on educational aspects. The faculty responsible for the genetic counseling courses at QU collaborates closely with the clinical team to ensure that the course content accurately reflects the realities of genetic counseling in Qatar. Guest lectures by experts from HMC and SM are incorporated into the program to provide students with the most up-to-date preparation for their clinical practice. Furthermore, expert faculty members from other QU departments, such as the Department of Psychology, contribute their specialized knowledge to enhance students' understanding of specific genetic counseling-related subjects. This approach demonstrates the program's commitment to staying connected with the evolving field of genetic counseling in Qatar.

The enrollment process for the program is an intricate and competitive undertaking. Prospective candidates undergo an online application process, wherein they submit their academic credentials and essential documents for admission. Based on the admission criteria,<sup>27</sup> applicants are then shortlisted for an interview conducted by selection committee members involving QU faculty and clinical faculty involved in the program.<sup>28</sup> This interview serves as a crucial evaluation phase, allowing the committee to gauge the candidates' suitability for the program.

The graduation process involves several key components. First, graduates must successfully complete the courses, as well as a professional development exam tailored specifically to assess their gained competencies through written and oral assessments. This exam serves as a pivotal evaluation, ensuring that graduates possess the necessary skills, knowledge, and ethical understanding essential for a successful career in genetic counseling. Students are also required to undertake a capstone project in genetic counseling, showcasing their ability to apply gained knowledge to develop relevant research questions and conduct high-quality and ethical investigations that may contribute to the existing knowledge in the field.

Given the program's commitment to maintaining high standards and ensuring optimal student-faculty ratios, especially during clinical training, the acceptance rate is currently limited to 3 to 5 students per year, ensuring a judicious selection process that prioritizes the most qualified individuals. Worth noting is that QU offers a competitive



graduate assistantship for nonworking students accepted for enrollment in QU graduate programs, including those in the genetic counseling master program, open for both local and international applicants. The program is a full-time program; however, students have the flexibility to complete the master's program in up to 4 years. Furthermore, QPHI started to sponsor students in the genetic counseling program, in 2022, as part of their commitment to building human capacity for the implementation of precision medicine in Qatar. Currently, 6 graduates from the master's program are actively contributing their skills and knowledge to the provision of genetic counseling services in the health care sector in Qatar, and 8 students are currently enrolled in the program. As the field continues to evolve, genetic counseling services are poised to broaden their scope, incorporating additional subspecialties such as cardiology and nephrology. The program in Qatar, represented by its Director, is currently a member of the board of directors of the Transnational Alliance for Genetic Counseling.<sup>29</sup> Transnational Alliance for Genetic Counseling comprises genetic counselor educators from over 20 nations, fostering connectivity among the global genetic counseling community and promoting enhanced international communication and collaboration.<sup>29</sup>

## Recognition of the profession

The Ministry of Public Health (MoPH) in Qatar has recently introduced national registration regulations for genetic counselors in Qatar, officially recognizing the genetic counseling profession and being the first among the Gulf Cooperation Council to have such recognition. According to these guidelines, genetic counselors are required to seek licensure from the Department of Health Care Professions at MoPH before practicing genetic counseling in Qatar.<sup>21</sup> To obtain an MoPH license, candidates must have a bachelor's degree in science or a health care field, a master's degree in genetic counseling with 2 years of clinical training in a recognized genetic counseling program, and board certification or eligibility from recognized credentialing bodies, although QU graduates are exempted from the board certification/eligibility requirement. Overseas candidates need a minimum of 2 years of work experience after certification or 4 years if board eligible, whereas specific exemptions apply for Qatari nationals, QU graduates, and offspring of Qatari women or residents, exempting these categories from the qualifying exam requirement. These ministerial guidelines specify the educational prerequisites for the profession, define the scope of practice for genetic counselors, and detail the criteria for competency validation and the renewal of licenses.<sup>21</sup> This achievement is also key for the protection and future development of the profession in the country.

## Research advancements

Qatar's commitment to research has led to the initiation of local genetic studies. These studies contribute to a deeper understanding of the genetic landscape specific to the Qatari population, allowing for more tailored and culturally sensitive genetic counseling services and paving the way for the implementation of precision medicine. The engagement in research reflects a forward-thinking approach, positioning Qatar as a hub for genetic advancements in the region.<sup>9</sup> Collaborative partnerships with experts from around the world have facilitated knowledge exchange, training opportunities, and the incorporation of best practices into the local context.<sup>9</sup>

Furthermore, Qatar stands out as one of the pioneering nations that have launched extensive population genomics initiatives aiming to facilitate precision medicine. This includes QBB and QGP, both which are currently under the umbrella of QPHI. QBB is an ambitious biobank that collects biological samples along with comprehensive biomedical test data, as well as comprehensive questionnaires, including health and lifestyle data, to enhance disease prevention, diagnosis, and treatment, as well as to explore the impact of lifestyle on disease development in Qatar.<sup>30</sup> In addition to QBB, QGP has a major contribution to genomic research in Qatar, by studying the genetic makeup of the Qatari population through sequencing the genetic map of the Qatari population toward the implementation of precision medicine and personalized health care. Through large-scale genomic research, educational initiatives, and partnerships, QGP has positioned Qatar as a global leader in advancing our understanding of genetics and its impact on the health care.<sup>31</sup> Hamad Bin Khalifa University in Qatar also offers a master's and a PhD degree in Genomics and Precision Medicine since 2017.<sup>32</sup> As the landscape of genetics and genomics research is expanding, the availability and role of genetic counseling in research entities are becoming more important.

Within the genetic counseling master's program curriculum at QU, students benefit from a research project component, many of such research projects are conducted in collaboration with HMC and SM faculty. Some of these studies were published in international journals,<sup>33,34</sup> whereas others are under preparation/review.<sup>35-38</sup> These research endeavors display rich diversity, investigating prospectively and/or retrospectively various conditions such as autism spectrum disorder (ASD), hearing loss, congenital heart diseases, retinal dystrophies, and various forms of cancer. They aim to elucidate the causative variants in the population of Qatar and the diagnostic yield of various genetic testing methods available for such conditions. By comprehensively analyzing these facets, the students seek to refine the genetic counseling approach for such conditions specifically for the Qatar population, contributing to a more

detailed understanding and tailored guidance. Simultaneously, other research projects focus on the knowledge, attitudes, and psychosocial aspects of genetic testing and counseling. One project aimed to translate and culturally adapt the Genetic Counseling Outcome Scale (GCOS-24)<sup>39</sup> for use in Qatar. Other ongoing research projects focus on investigating the outcome of national screening programs, such as premarital screening and NBS, in addition to testing methods such as prenatal ES. By addressing these diverse contexts, the students aim to fill critical gaps in genetic counseling. This multifaceted research approach underscores the commitment of students and their supervisory team to advancing the field of genetic counseling in Qatar and the region, ensuring a comprehensive and well-informed framework for addressing a wide array of genetic and hereditary conditions.

### **Challenges and ethical considerations facing the genetic counseling profession in Qatar**

One of the challenges facing the genetic counseling profession in Qatar is the limited awareness and understanding of the profession itself and the genetic services available in Qatar among the general population, as well as some misconceptions related to genetic testing that may affect the public attitude toward genetic testing and counseling. To overcome such challenges, strategies such as increasing public awareness through educational campaigns, academic programs, media platforms, and religious scholars' involvement can be implemented, taking into account the cultural and religious factors affecting public knowledge and attitude.

One notable area for improvement is to have more integration of genetic counselors with all subspecialties. In Qatar, genetic counselors often work in isolation from the rest of the care team, and the necessary multidisciplinary team clinics and connections are not fully established. Although genetic counselors receive referrals from various subspecialties and provide valuable services, there is a need for enhanced collaboration and integration. Establishing focal points, discussing cases, and fostering close collaboration between genetic counselors and other specialists are crucial steps to ensure they are fully utilized. For instance, at HMC, a close collaboration between genetic counselors and the Adult Nephrology-Kidney Transplant Team has been established, agreeing on approaches to genetic testing for their patients. Such collaborative efforts should be expanded across all subspecialties to maximize the effectiveness of genetic counseling services.

In addition, referring patients to genetic counseling services needs improvement to optimize the health system's performance in preventive and precision medicine. Under-referral denies eligible patients access to genetic counseling services, whereas premature or inappropriate referrals that do not meet referral criteria burden the system. Solutions include standardized referral criteria,

physician education, and improved communication channels. A promising future focus is to educate nongenetic health care professionals in subspecialties such as cardiology, nephrology, and oncology to order genetic testing. This strategy enables genetic counselors to concentrate on posttest counseling, tailored management, and continuity of care. By integrating genetic education across medical fields, we can optimize the use of genetic counselors and improve the overall quality of patient care. Additionally, evaluating the effectiveness of international guidelines versus local ones is essential, considering social, religious, and ancestral factors. Tailoring genetic counseling to cultural sensitivities, investing in community outreach, and ensuring equitable access can effectively address these challenges.

The high inbreeding and consanguinity rate in the Qatari population, as well as the large extended family structures, place a significant demand on genetic counselors. Additionally, polygamy is occasionally pursued to mitigate genetic disease recurrence. Cultural beliefs, notably regarding the "evil eye," often lead families to attribute genetic disorders to supernatural causes, affecting their willingness to undergo genetic testing and preventive measures. The importance of cultural competence in genetic counseling cannot be overstated. In a highly diverse population such as Qatar, it is a challenge for genetic counselors to navigate diverse cultural backgrounds, considering the impact of cultural and religious beliefs and values on individuals' perceptions of genetic information.<sup>21</sup> Tailoring counseling approaches to respect cultural diversity is integral to establishing effective communication and ensuring that individuals feel heard and understood along with their psychological and emotional needs. Thus, one possible aspect for improvement could be having genetic counselors from more diverse backgrounds that can provide counseling in various groups and perhaps languages. A significant challenge faced by genetic counselors in clinics is the large proportion of Arabic-speaking patients. This often necessitates that genetic counselors speak Arabic to effectively communicate with these patients. When this is not possible, reliance on medical interpreters becomes essential. Although interpreters are invaluable, their use adds a layer of complexity to the genetic counseling process. Interpreters must accurately convey complex medical terminology and navigate cultural nuances, all while ensuring confidentiality and mitigating accuracy concerns. This can be challenging and time consuming, highlighting the need for genetic counselors to be proficient in Arabic or to have reliable and skilled interpreters readily available.

In relation to ethical considerations, genetic counseling in Qatar involves navigating multiple ethical and culturally sensitive considerations. In this predominantly Muslim country, religious beliefs often play a significant role in influencing patients' decisions. For instance, the topic of termination of pregnancy once a challenging subject for discussion, has evolved over time. The acceptance of termination of pregnancy in Qatar is contingent on specific

circumstances, such as endangerment to the mother's life or the presence of a life-limiting condition in the fetus.<sup>40</sup> According to Islamic and legal laws in the country, these decisions must be made before 120 days of gestation.<sup>40</sup> Genetic counselors approach these discussions with utmost respect for patients' choices, taking into account their cultural and religious beliefs.

Moreover, privacy and social stigma concerns pose another cultural dilemma in Qatar. Many families hesitate to engage with genetic counselors because of fears that seeking genetic services might label their family names with a genetic condition, potentially affecting their reputation and leading to social stigmatization.<sup>21</sup> Genetic counselors address these concerns by assuring patients that their genetic data are securely stored and not shared with anyone. This emphasis on confidentiality aims to build trust and encourage patients to open up about their genetic concerns, fostering a more supportive and understanding genetic counseling environment.

Furthermore, the cultural and religious attributes inherent to Qatar and the broader Arab/Muslim world necessitate the presence of evidence-based genetic counseling expertise that is custom tailored to suit the needs of these societies. QU's Genetic Counseling master's program is designed to integrate this specialized knowledge, ensuring that future practicing genetic counselors in the country possess the necessary competence in addressing these unique aspects.

## Conclusion

In conclusion, Qatar's genetic and genetic counseling landscape has evolved significantly during the last 2 decades, driven by its unique demographics and cultural nuances. This evolution has been supported by proactive initiatives and institutions and dedicated educational programs. This has led to the establishment of comprehensive genetic counseling services for prenatal, pediatric, and adult cancer, as well as serving important national screening programs like premarital, newborns and breast cancer screening programs. The recognition of the genetic counseling profession is marked as a significant achievement for Qatar toward the protection and future expansion of the profession.

In addition, the introduction of the genetic counseling master program at QU demonstrates Qatar's commitment to fostering the indigenous expertise of competent and culturally sensitive genetic counselors who can serve its population and shape the future of personalized medicine in Qatar. The expansion of genetic counseling services, with a focus on subspecialties, will further cater to diverse health care needs.

Concurrently, ongoing research initiatives in genomics pave the way for precision medicine implementation in Qatar. Looking ahead, Qatar's genetic counseling future holds promise, driven by advancements in genetic testing, research endeavors, and educational initiatives.

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## Conflict of Interest

The authors declare no conflicts of interest.

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