

# Genetics and Genomics Nursing in Saudi Arabia: Opportunities and Challenge

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## Abstract

Nurses are critical members of the health-care team, and with sufficient knowledge and training in genetics and genomics, they can contribute significantly to fulfilling the potential of precision medicine in practice and research. The efforts of Saudi Arabia toward genetics and genomics are remarkable. Yet, the voice of nurses in such an important field has not been sufficiently explored. Several potential challenges in the health-care system related to genetics and genomics nursing must be considered. In this article, we outline the challenges and opportunities of nursing genetics and genomics fields in Saudi Arabia.

**Key words:** Genetics, genomics, nursing, Saudi Arabia

## BACKGROUND

The rapid growth of genetics and genomics science has substantially changed the approaches to screening, diagnosing, treating, preventing disease, and monitoring therapeutic success in healthcare.<sup>[1]</sup> This growth of genetics and genomics has resulted in an expanded role of health-care providers, including nurses. Genetics has been defined as the “study of genes and their roles in inheritance of disease.”<sup>[2]</sup> While genomics is the study of the entirety of a human’s genes.<sup>[3]</sup> Due to their widespread availability, nurses can make a big contribution to preventing and assisting people affected by genetic disorders.<sup>[4]</sup> Therefore, they need to be equipped with knowledge and skills to apply genetic advancements,<sup>[5]</sup> particularly with the steadily increasing use of genetics and its implications in the health-care sector.<sup>[6]</sup> According to the American Nurse Association (ANA) and International Society of Nurses in Genetics (ISONG), genetics/genomics nursing is the “protection, promotion, and optimization of health and abilities, prevention of illness and injury, alleviation of suffering through the diagnosis of human response, and advocacy in the care of the genetic and genomic health of individuals, families, communities, and populations.”<sup>[7]</sup> Genetic information is used for disease diagnosis to detect heritable changes or variations in gene sequences or chromosome structures.<sup>[8]</sup> Genetics is critical to the role of nurses in risk assessment, analyzing the genetic

component of cancer, heart disease, diabetes, and Alzheimer’s risks, and discussing the impact of risk on individual and family health-care management.<sup>[9]</sup> However, globally, nurses lack sufficient knowledge and competence in the field of genetics,<sup>[10]</sup> especially with lacking educational guidelines, framework, and scope of practice for nursing genetics.<sup>[11,12]</sup>

## GENETICS IN NURSING

Nurses are required to be capable of conducting comprehensive and holistic assessments of their patients to lay the groundwork for patient care. Family history is a critical component of nursing assessment, which is a required skill for all nurses. It is the critical criterion for identifying individuals who may benefit from genetic counseling. Therefore, it is important to prepare nurses to incorporate genomics into routine clinical practice.<sup>[13]</sup> Nurses have been incorporating genetics into their practice since 1970s.<sup>[4]</sup> Worldwide, many nursing professional organizations have been formed to aid nurses and other healthcare workers in comprehending and applying genetic discoveries to patient care in daily practice.<sup>[14]</sup> The ISONG is a well-known society that is tasked with the responsibility

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of supporting scientific and professional progress in the discovery, interpretation, application, and management of genomic information. The ISONG sets scope and standards of practice for genetics and genomics nursing, incorporating them with the ANA.<sup>[7]</sup> Nurses work in many countries as part of the genetic multidisciplinary team, such as Japan, the US, Australia, Canada, Germany, and Hong Kong.<sup>[11]</sup> Genetic nurses are contributing significantly to the care of families affected by familial cancer, working as genetic counselors, caring for patients with genetic diseases and congenital disorders, setting educational objectives for genetic nurse education and training, delivering education to patients and families, administering prescribed medication, advocating for disease prevention and health promotion, collecting and interpreting family history information, and partnering with the health-care team to facilitate genetic referrals as needed.<sup>[15-17]</sup> However, the primary challenges identified across countries toward nursing genetics included defining the role of the specialist nurse in genetics, developing genetic nursing competencies, inadequate attention to genetics as a nursing issue, lack of genomic literacy among nurses, and a lack of evolving genetics/genomics in nursing curricula.<sup>[11]</sup>

Efforts have been made in the US to provide nurses with genetic and genomic competencies as part of their job. As a result, the *Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines, and Outcome Indicators* were released to specify the essential competencies for all registered nurses in genetics and genomics.<sup>[18]</sup> In the UK, genetics as a specialization in nursing was acknowledged in the late 1970s by establishing the Genetic Nurses and Social Workers Association, and advocate nurses as a vital part of the genetics team, to promote their professional development in their role. However, their roles were limited to providing psychosocial support to the families. Then, in the middle of the 1990s, genetic nurses took on more responsibility for their caseloads and gained more independence in the clinical genetics team.<sup>[16]</sup>

Genetics literacy and competency were examined thoroughly in nursing research by utilizing the Genetics and Genomics Nursing Practice Survey by Calzone *et al.* and the Genomic Nursing Concept Inventory by Ward (2011).<sup>[10,11]</sup> The results of both instruments demonstrated the literacy of genetics was low and there is a need to expand the knowledge of genomics and genetics across diverse samples, including registered nurses nursing students, and faculty.<sup>[19-21]</sup> To the best of our knowledge, there is no such study assessing nursing genetics literacy in Saudi Arabia.

Nurses are critical members of the health-care team, and with sufficient knowledge and training in genetics and genomics, they can contribute significantly to fulfilling the potential of precision medicine in practice and research.<sup>[22]</sup> According to the American Association of Colleges of Nursing,<sup>[23]</sup> the important domains of nursing education in genetics and genomics include professional responsibility, professional

practice, and the provision of education, care, and support. In genetic clinical settings, nurses' roles are about obtaining a family history, delivering information about genetic tests, including their duration and risks, offering counseling and psychological support, identifying hereditary risks, understanding monogenic and multifactorial disorders, and interpreting genetic test results.<sup>[24,25]</sup>

Nurses must increase their genetic expertise as part of interdisciplinary teams. Strategies to inform the nursing community about adopting genomics into practice include developing a network of genomic experts as points of contact.<sup>[26]</sup>

## GENETICS IN SAUDI ARABIA

The Kingdom of Saudi Arabia is the largest country in the Arabian Peninsula with a current population of 35,728,65 million. Saudi healthcare is undergoing reform in line with the Saudi 2030 vision to create a comprehensive, effective, and integrated health system centered on the individual and society's health, with a focus on enhancing the quality and efficiency of healthcare and promoting the prevention of health risks. In the 2030 vision, genetics was given significant attention. The Saudi Genome program was developed, which aims to reduce the number of genetic diseases in the kingdom, set up a genetic database, and document the first Saudi genetic map through the sequencing of the Saudi population's genome.

The history of genetics in Saudi Arabia started in 1978 when King Faisal Specialist Hospital and Research Center (KFSHRC) hired Dr. Nadia Sakati to be the first clinical geneticist in Saudi Arabia. She co-founded genetic services at KFSHRC with Prof. Pinar Ozand.<sup>[27]</sup> The first Saudi genetic counseling postgraduate diploma was established in 2005 in KFSHRC, with five graduated genetic counselors.<sup>[28]</sup> In 2014, with the collaboration of Alfaisal University, the program expanded into a master's degree in Genetic Counseling. Moreover, the medical genetics program was founded in 2011 to identify and treat a variety of genetic abnormalities, provide genetic counseling, and treat lysosomal storage disorders by stem cell transplantation. Numerous hospitals provide clinical genetic services, such as King Fahad National Guard Hospital, King Fahad Medical City, King Saud Medical City, King Saud bin Abdul-Aziz University, King Abdul-Aziz Medical City, King Abdallah International Medical Research Center, King Khalid University Hospital, and King Abdallah University Hospital.<sup>[27]</sup>

The efforts of Saudi Arabia toward genetics are remarkable. As an example, the first Saudi national newborn screening program for congenital hypothyroidism began as a pilot study in December 1988 and was expanded to encompass all deliveries at the Ministry of Health and King Saud University hospitals by the end of 1990.<sup>[29]</sup> Moreover,

since the prevalence rate of  $\beta$ -thalassemia and Sickle Cell Disease in Saudi Arabia is the highest in the Middle East (0.05% and 4.50%, respectively), the National Premarital Screening Program for sickle cell disease and thalassemia was established in 2001 and made it a requirement before acquiring a marriage license in 2004.<sup>[30]</sup>

Several genetics educational programs are offered in Saudi Arabia, including fellowship programs for clinicians at King Faisal Specialist Hospital and Research Center, King Fahad Medical City, and King Fahad National Guard Hospital. Master's degrees for genetics counselors at Alfaisal University and King Abdul-Aziz University. Master program for laboratory personnel at King Abdul-Aziz University.<sup>[27]</sup> At present, the only academic course that is offered for nursing students in genetics and genomics is an elective course in the Ph.D. nursing program at King Saud University.

## OPPORTUNITIES AND CHALLENGES TOWARD NURSING GENETICS IN SAUDI ARABIA

The concept of consanguineous marriage is popular in Saudi Arabia and other Middle Eastern countries, with is estimated to be 56% in Saudi Arabia<sup>[31]</sup> and 39.8% in Riyadh's capital city.<sup>[32]</sup> Which explains the increased number of Autosomal recessive conditions.<sup>[33]</sup> According to the Catalogue of Transmission Genetics in the Arabs database, 67.3% of diseases recorded in Saudi Arabia have recessive inheritance.<sup>[34]</sup> Hematology, endocrine, and metabolic disorders are the top three prevalent pathogenic variations carried out among the Saudi population.<sup>[35]</sup> The high prevalence of genetic disorders in Saudi Arabia fosters the need for a competent health professional who is capable of collecting and evaluating family and medical history, conducting a risk assessment, educating patients about genetic testing, acquiring informed consent, and providing psychological assessment and support. Nurses are uniquely qualified to deliver genetic health information, assessments, education, and counseling. In addition, genetic counseling falls within the scope of their nursing practice.<sup>[2]</sup>

Several potential challenges in the health-care system related to genetic nursing must be considered in Saudi Arabia. Even with the availability of postgraduate genetics counseling programs, the issue of credentialing and role recognition from the Saudi Commission for Health Specialties and other legislators hindered nurses' efforts to develop a genetics solid background. Instead of the role of advanced practice nurses (APNs) in genetics and genomics to provide genetic counseling, order and interpret genetic testing, and monitor and manage individuals affected by or at risk for genetic diseases,<sup>[36-40]</sup> the scope of service of APN is impeded by a variety of legislative and practical constraints. The definition of core competencies for genetic/genomics nursing and

related genetics knowledge is not defined in Saudi Arabia, which may be due to the policymakers' failure to recognize nurses as stakeholders in the provision of genetic/genomics healthcare. The other challenge concerned the absence of an educational strategy to integrate genetics into undergraduate nursing programs. In addition, the lack of nursing genetics expertise to teach genetics has an impact on the awareness and literacy of nurses.

A well-structured action plan is required to overcome the current challenges. Legislators, committees, and others must be involved in the areas of action. Formal nursing committees must advise and encourage the Saudi Commission for Health Specialties to assist nurses' registration and accreditation as genetic counselors. APNs must be encouraged and supported to develop genetics skills within a defined scope of practice. Integrating genetics and genomics into the nursing curriculum, as a core course is required. Through teaching and training, we raise awareness and comprehension of the importance of genetics in nursing practice. In addition, establishing collaborations with international nursing networks and organizations in the UK, US, and Japan, such as ISONG, will help the Saudi nursing workforce comprehend the function of the genetic nurse in practice. Policymakers are advised to develop a policy that requires nurses to include genetics in their practice. Finally, future nursing research on genetics is necessitated to figure out the current situation and come up with an action plan to close the gaps.

## CONCLUSION

Through their role as advocates, nurses are well placed to play a major role within the Saudi health-care workforce by integrating genetics into their clinical practice. Addressing current and future challenges in clinical, education, policy, and research is recommended to provide a solution to these challenges and foster the nursing role and importance in the field of genes.

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