Genetics Counseling in Saudi Arabia

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Abstract: Genetic counseling is "the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease." This process includes collecting and interpreting the family and medical history, risk assessment, a comprehensive educational process for potential genetic testing, informed consent, and psychosocial assessment and support. The aims of this review is to spot the light on genetic counseling services in Saudi Arabia, exploring the cultural perspective and attitude toward genetic counseling, investigate factors hinder genetic counseling and discuss the nurses role in genetic counseling. Genetic counseling started in Saudi Arabia in 2005 where the first Saudi genetic counseling training program was established by the Department of Medical Genetics at King Faisal Specialist Hospital and Research Center (KFSH&RC) and considered as a major referral centre in the Kingdom of Saudi Arabia. (KFSH&RC) initiated the first metabolic clinic ².

I. Introduction

Genetics is the science of inheritance. The word Genetics is derived from ancient Greek word "genitive" that means "origin". It aims to appreciate the mechanism by which the blueprints for life are passed through generations and variations in these blueprints are vital for evolution ³. The contemporary science of genetics, seeks to comprehend the process of legacy, only began with the work of Gregory Mendel in the midnineteenth century and he observed that organisms become heir to traits via discrete units of inheritance, which are now called genes.

Genetic counseling is "the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease." This process includes collecting and interpreting the family and medical history, risk assessment, a comprehensive educational process for potential genetic testing, informed consent, and psychosocial assessment and support. A cornerstone of a genetic assessment is obtaining and interpreting the family history, whether by phone, through a paper or webbased interface, or as part of a clinic visit. Traditionally, the personal and family medical histories have been used to develop a differential diagnosis, to identify and quantify risk for family members, and to select the appropriate test (Pyeritz 2012) 4.

Marriage between cousins has been part of the culture for millennia leading to "founder" effect and a large number of autosomal recessive diseases ⁵. Available data suggest that genetic and congenital disorders are more common in Middle Eastern countries; Several factors may contribute to the high prevalence of genetically determined disorders: The high prevalence of haemoglobinopathies, glucose-6-phosphate dehydrogenase deficiency, autosomal recessive syndromes, and several metabolic disorders ⁶. The rate of children with Down's syndrome in some Middle Eastern countries exceeds the 1.2-1.7 per 1000 typical for industrialized countries. This may be related to the relatively high proportion of births to older mothers in the region (up to 50% of children with Down's syndrome in the region are estimated to be born to mothers aged 40 or over) ⁷.

The Kingdom of Saudi Arabia (KSA) is an Arab Middle Eastern country that lies at the furthermost part of southwestern Asia. It occupies four/fifths of the Arabian Peninsula. The gene for sickle cell disease was first discovered in the Eastern Province of the Kingdom of Saudi Arabia (KSA) in 1963. Consequently, a series of investigations has revealed beside the sickle cell gene, the presence of thalassemias and red cell enzymopathies ^{8,9,10}. The population according to the 2013 census is approximately 29,9 million people, among which around 9 million are non nationals (**The Saudi Central Department of Statistics and Information** 20103). The prevalence of genetic disorders in Saudi Arabia was determined to be 1,138,065 with a rate of 3.7 percent of births. This rate is higher than the USA rate (2.5\$) ¹¹.

Genetic counseling is nondirective. This means that counselors provide the family with all the available options free from the bias of an individual counselor or from preconceived ideas the counselor may have about the family. **Phillips (2001)** ¹². Genetic counseling provides an individual or family with information and support regarding health concerns which run in their family. Genetic counseling may involve the diagnosis of a genetic condition, the provision of information and supportive counseling (advice and guidance) by a team of health professionals, so that families and individuals may be better able to adjust to diagnosis. Follow-up counseling is available to ensure on-going support, review previous information or to answer new questions as they arise ¹³. Genetic counseling can assist women or couples who are:

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- Planning a pregnancy
- Interested in prenatal diagnosis
- Concerned about first or second trimester screening results
- Known to be at risk for carrying genetic disorders such as cystic fibrosis, muscular dystrophy, hemophilia, sickle cell disease or thalassemia
- Pregnant and will be 35 years or older at the time of delivery
- Of an ethnic group in which particular inherited diseases are more common, including African-American, French-Canadian, Mediterranean or Asian backgrounds

II. Genetic Counseling Services

Saudi Arabia has a high prevalence of genetic diseases as proven by several studies led by KFSH&RC and other medical centers from around the Kingdom. This high incidence is due to the high rate of consanguinity and rapid population growth. This drastic rise in the incidence of genetic disorders among the population created an urge to have compulsory premarital testing for genetic diseases ². The incidence of many autosomal recessive genetic disorders in Saudi Arabia is high when compared to other populations (AL-Owain et al. 2012) ¹⁴.

In Aug 2005 the National Newborn Screening Program was introduced in the Kingdom. The program has graduated five genetic counselors with high diploma-level degree ¹⁵.

Two programs have been adopted at the national level; compulsory premarital testing for Sickle Cell Anemia and Beta-Thalassemia and Newborn screening for 16 inherited metabolic & endocrine disorders. However, strategies need to be established to ensure success of such programs by constant revision and troubleshooting ¹⁶.

In Saudi Arabia, Under the Royal decree, of 4/1/1423 H (i.e. 8/3/2002 G) Ministry of Health in Saudi Arabia made organizational arrangements for premarital screening program. And the premarital screening started on 1/1/1425 under a second Royal decree, which made premarital screening for genetic diseases mandatory for all those couples who plan to marry and the marriage contract were not be issued until result of this screening test was submitted. However the couples had the choice of getting married, even in case of incompatible results¹⁵. The Saudi Ministry of Health designed a protocol for premarital testing after the Royal Decree in December 2003 and implemented in February 2004. From February 2004 to February 2008, 60,000 healthy subjects coming to six marriages consultation centers in the Al-Hassa area underwent routine mandatory tests. At the beginning, Saudi premarital screening program only covered Sickle cell disease and Thalassemia. At present a network of 122 reception centers, 70 labs and 20 genetic counseling clinics had been involved in the screening program ¹⁷.

Natural Working Group (NWG) was established to coordinate various activities and services. This group, a voluntary body, was established in 1990 and a National Committee at the Ministry of Health was established in 1994. Both integrated expertise working in relevant fields and both worked closely to standardize services and various parts of the country. The NWG incorporates members from various disciplines including physicians, nurses, laboratory specialists, social worker, family members of affected individuals and advocates from the community. The coordinators and deputy coordinate were chosen from various parts of the country. To plan and implement the suggestions and views of the NWG, a National Committee was appointed by the Minister of Health. The national coordinator is the chairman of the National committee for blood genetic disorders at the Ministry of Health with membership from other health institutions. The WHO Collaborating Center (WHOcc) was established in 1992, where Department of Biochemistry was designated as the "Collaborating Center for hemoglobinopathies, thalassemias and enzymopathies with specific tasks to be carried out at National and Regional Levels ¹⁸.

The main tasks of the national working group include the following: To assess and update the frequency and distribution of Saudi population with genetic disorders in the Kingdom of Saudi Arabia, improve management and care for patients suffering from genetic disorders, increase awareness and educational program of relevance, implement the national control and prevention strategies, conduct research and involve in local, national, regional and international peer links plus interactions.¹⁸

The major objectives of national committee for genetic disorders are: National Committee coordinates the activity and hold a yearly meeting to discuss new development, reveal on the achievement of the last year and predict the projected plan for the following year. Assessment, formation and evaluation of epidemiological situation, care policies related to genetic diseases, preparing a national program for control and prevention of genetic diseases, adopting and coordinating quality control. ¹⁵

The main tasks of WHO collaborating center include: Community based services for the genetic disorders, community awareness regarding hereditary genetic diseases, training for the population in the fields of investigations, diagnosis and care for hereditary diseases, participate in the implementation of inter and intra-

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country meeting recommendations and regional programs on prevention and control of the hereditary diseases, conduct research in genetic services and ethical aspects in the relation to the Islamic believes and traditions ¹⁹.

Screening milestones 20

- 1991 Cord blood Hypothyroidism screening at 20 centers
- 1994 Introduction of tandem mass spectrometry at KFSH&RC
- 1995-1998 First MS/MS pilot study, published 1999
- 1995-2005 Expanded newborn screening on small scale
- Aug 2005 National newborn screening started
- Phase I includes 24 birth centers and 120,000 newborns
- 2008 Coverage of >400,00 newborns

The genetic counseling program in KFSH&RC was accredited by the Saudi Commission as a postgraduate course. It is a high diploma-level program with duration of 2 years and a 6-month clinical internship. Presently the total number of genetic counselors in Saudi Arabia is ten where nine of them are Saudis. Four of them graduated with a Master's-level degree and one (non-Saudi) graduated with a Master's-level degree. The remaining five graduated with a high diploma-level degree from the genetic counseling training program at KFSH&RC ¹⁴.

Genetic counselors enter the field from numerous disciplines including the fields of biology, nursing, psychology, public health and social work. Most counselors graduate from accredited programmers in genetics or genetic counseling. Currently there are a total of 26 training programmers accredited by the American Board of Medical Genetics. Twenty-two are in the USA, two in Canada, one in the UK and one in South Africa. These programmers emphasize basic science concepts in medical, biochemical and molecular genetics as well as statistical analysis, and psychosocial counseling skills. Each programmed accepts approximately four to ten candidates each year, with the number of new graduates exceeding 100 ¹¹.

Graduates earn a Master of Science degree and are encouraged to sit for board examinations administered by the American Board of Genetic Counseling. The National Society of Genetic Counselors boasts 1500 members from many different countries. Employers include university-based programmers, private hospitals, prepaid health plans, diagnostic laboratories and government- supported health agencies. These counselors often specialize in particular areas. More than half work in prenatal diagnosis' Approximately 20% work in a pediatric setting. Only 1% specializes in adult-onset genetics. The fastest growing group, cancer genetics counselors, comprises almost 10% of all board-certified counselors.

In some short year's genetic nursing practice has been transformed from a nearly concealed specialty to a recognized specialty practice with formal recognition, publication of scope and standards of practice, and most recently the availability of credentialing for genetic nurses.

The influence of recent genetic advances on nursing practice is especially evident in oncology. Oncology nurses practicing in cancer prevention and control apply genetic principles to their clinical practice daily. "Genetic nursing is a holistic practice that includes assessing, diagnosis, planning, implementing, and evaluating the physical, spiritual, ethical, and psychosocial aspects of patients and families who have genetic concerns". In addition, advanced practice nurse (APNs) in genetics provide genetic counseling, order and interpret genetic tests if within their scope of practice, and provide surveillance and management of persons affected by or at risk for genetic conditions ²¹.

III. Attitude of Saudi toward genetic counseling

One of the major challenges in the field of genetics and disease prevention is the outlook of Saudi couples toward reproductive decisions. Many couples decide to go ahead with the marriage despite the result that they are carriers for genetic disorder. This may be due to lack of understanding or ineffective genetic counseling ¹⁵. The effectiveness of carrier screening depends largely on the knowledge and attitudes of the target population. Also, there are specific factors (gender, area of upbringing and type of education) affecting the attitudes toward genetic counseling. Some studies investigate the degree of knowledge regarding genetic disorders and the national premarital screening program in an important subgroup of the population (young university undergraduate men and women in the city of Jeddah). The studies showed that despite a reasonable level of knowledge regarding genetic disorders in general, there was a lack of appropriate knowledge regarding the national premarital screening program.

Genetic disorders cause a significant burden in Saudi Arabia, and hemoglobinopathies make up a substantial proportion of these genetic disorders. Screening for the carrier status may help in decreasing the occurrence. The local attitudes towards the screening program are an important determinant of its success rate. A study done to investigate knowledge regarding the national premarital screening program among university students in western Saudi Arabia, found that medical students consistently had more knowledge than non-

medical students and women are more knowledge than men' Also, women had stronger attitudes toward the accomplishment of testing with a significantly higher number of female respondents believing that the premarital screening should be mandatory and that marriage should not be allowed between two carriers of the same disorder.²²

A study done by **Al-Aama**, (2010), investigate the attitudes towards mandatory national premarital screening for hereditary hemolytic disorders. The author concluded that above 60% of the sample agreed that marriage should be prevented. Although this represents the majority, there is a significant proportion that does not agree. This group must be kept in the mind of policy makers. Rather than preventing the marriage which may cause significant social and individual harm, it would be better off looking at the reasons why couples often decide to marry despite both members being carriers of the same disorder. One very important factor is the timing of the test. ²³

Moreover, a study done to investigate the perception of the female students of King Saud University on the application of premarital screening found that 75 % supported the compulsory application of premarital screening. The study concluded that pre-marital screening could be extended to include a broader spectrum of health/genetic disorders and will be useful for early identification and possible intervention as well as the prevention of complications.²³.

In recent years, premarital counseling has gained acceptance. A study conducted among the general population in Riyadh in 2008 showed a positive attitude toward the program and that the majority of participants agreed with the idea of applying the PMSGC program to all couples in all regions of the country ²⁴.

Another study conducted by Al-Khaldi, etal. (2010) found that 70% of the students showed acceptance of premarital counseling .Family history and blood tests for hemoglobinopathies were the most common 2 items preferred to be included in premarital counseling by the respondents.²⁵

Genetic counseling followed by testing should be done in group sessions in the early college years or on leaving school with parental consent for those under 18 years of age. It would be essential however to have counseling appropriate to each individual's age and understanding so as to prevent unwanted complications of genetic testing such as anxiety, social stigmatization and discrimination. Genetic counseling can be followed by testing if the individual decides to do so. Also, improving the population knowledge will improve their understanding and cooperation and can decrease the number of marriages among carriers without resorting to legal restrictions.

Public Education regarding genetic disorders and the implication of testing is essential for the success of the premarital screening. In addition to formal counseling at the time of testing, mass education especially at high school and university levels can play a major role in increasing the level of knowledge regarding premarital screening and genetic counseling (PMSGC). It has also been suggested that education about pre-marital screening be included in the university curriculum. Opportunistic counseling done by health professionals during general check-ups or other physician office visits may help in addition to educational pamphlets, targeted television programs and advertisement. Perhaps education must start with those in the medical profession especially those in certain specialties such as family medicine or obstetrics and gynecology so they can play their role in the education process and be equipped to answer related questions.

IV. Role of nurse in genetic counseling

Nurses specializing in maternal healthcare play a key role in the prevention of genetic diseases. The amount of involvement depends on the nurse's education, understanding of genetics, job description and the type of genetic disorder the client has. Some nurses may specialize in genetics and become genetic counselors. But most nurses are in a liaison position with more specialized personnel .By accruing facts of genetic disorders and using their powers of observation, nurses may be the first to discover 'clues' and recognize a genetic problem and endow with referral services . Nurses should be alert to the heritability of many disease entities, and should take responsibility for helping patients to obtain a genetic referral. They should inform all women aged 35 or older of their increased risk of giving birth to a child with chromosomal abnormalities, and of the availability of prenatal diagnosis. Genetic nursing practice includes but is not limited to the following activities: client and family assessment to identify genetic risk factors and intervention, information, service, and referral needs, take a detailed family history and construct a pedigree, analyze the assessment data, provide genetic education, and develop and carry out a plan of care to address genetic concerns ²⁶.

Assessment

A thorough pre-conception history identifies couples who are genetically at risk. Women of childbearing age and their partners should always be screened for a genetic history. When women and their partners are informed of the risks of having a baby with birth defects or a genetic disorder prior to pregnancy, they are then able to determine their options regarding a pregnancy (including contraception, artificial insemination, adoption, prenatal invasive testing, or chance) ²⁷. The nurse should be able to make a referral to

an obstetric or genetic service where prenatal diagnosis and amniocentesis are available. A federal programmed on genetic counseling throughout US has been in effect since 1978 ²⁸.

Interviewing skills are extremely important in obtaining the relevant preliminary genetic history. The experienced nurse may also chart the family's pedigree in preparation for genetic counseling. The nurse should collect history about family dynamics. In many cases the family medical records will be needed for further research into the occurrence of a disorder in the family. Sometime some clients don't want others in the family to know that they are having familial defects. They want to keep it secret. The matter of obtaining permission to see family records must be handled with utmost delicacy and in a non-threatening way. The nurse may prepare or help the pregnant woman prepare for a diagnostic test both by discussing the tests, the procedure, the possible outcomes and the length of time needed for the test and results, and by assisting with procedures in the physician's office, the clinic or the hospital. After taking history and preparing the client for the referral sessions, the nurse should caution that client or family against expecting immediate results. They may tell that it may take time to get the result or answer from the counselor.

V. Diagnosis

The nurse may assist in preparing family members for specific genetic diagnostic testing and counseling. Some nursing or collaborative the problems should be identified by the nurse. The following are some nursing diagnoses that may be particularly useful in the care of families with genetic problems: Decisional conflict, Grieving, Knowledge deficit, Disturbance in self-concept

Planning and implementation

Planning and implementing follow-up is probably the most important care the nurse can give a couple undergoing genetic counseling.

Interpretation and reinforcement: The nurse is required to interpret and emphasize information given by the geneticist. The nurse should be able to answer further questions, clarify the information and discuss management facilities, community agencies for support.

Assistance for decision-making: Support should be given both in making decision and after the decision has been made. Decisions will be emotionally painful. They may involve the birth of a defective baby or such things as termination of pregnancy, sterilization, adoption or artificial insemination. The nurse should give support and make arrangements to facilitate decisions.

Evaluation

Once genetic care is indicated the nurse should evaluate the adequacy of patient teaching and supportive care on an ongoing basis. Finally when families are faced with difficult decisions with respect to genetic outcomes, the nurse is responsible for ongoing follow-up to ensure that adequate information and adequate anticipatory guidance is available ²⁹.

VI. An innovative landmark in Genetics Nursing Practice

Genetics nursing practice has seen numerous exceptional milestones. In the past, there has been minimal recognition of the important role that nurses can play in genetics. The International Society of Nurses in Genetics (ISONG) has undoubtedly been the leader in functioning with nursing leaders to endorse genetic nursing practice and build up a credentialing training for genetic nurses.

Through these efforts, only five years ago the American Nurses Association established genetics nursing as an official specialty of nursing practice. This statement was followed by the publication of the scope and standards of clinical genetics nursing practice by the American Nurses Association (ANA) and the International Society of Nurses in Genetics (ISONG) (11). Although determining credentialing standards is not an easy process, ISONG recognized that genetic nurses needed to have credentialing to establish competency, and a credentialing committee was formed in 1999 ³⁰.

Obstetric nurse can play a key role in the prevention of genetic diseases by learning the indications for patient referral for prenatal testing. It is extremely important that the nurse use her interviewing skills to obtain a relevant preliminary genetic history so that pertinent family and medical history can be evaluated and appropriate referrals made. These efforts will help to ensure that all patients, regardless of socioeconomic status, have access to genetic service

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