Premarital testing Screening in KSA

The inherited haemoglobinopathies, a group of disorders that include thalassaemias and sickle-cell disease, are a major public health problem in the Mediterranean region, the Middle East, Indian subcontinent, Asia, tropical Africa and the Caribbean. However, because of population flow, they are now widespread and occur in Europe and North and South America. According to the World Health Organization (WHO), approximate estimates of affected individuals indicate that 240 million people are heterozygous for these disorders and at least 200,000 lethally affected homozygotes are born annually, approximately equally divided between sickle-cell anemia and thalassaemia.[1] As consanguineous marriage is common in most Arabic-speaking countries, the incidence of such diseases is high, such that 5%-10% of Arabs carry the thalassaemia gene.[2,3]

The WHO has recommended several measures for the prevention of genetic diseases, such as health education, screening to identify individuals or couples at risk, genetic counseling and prenatal diagnosis. For these aspects of prevention to be applied to a population, various ethical, legal, and cultural issues have to be taken into account. Any preventive campaign must therefore be tailored to the needs of each culture.[1]

Genetic counseling is a new field of medicine demanding a comprehensive knowledge of genetics and the management of genetic disease, as well as its impact on the individual, the family, offspring and the community at large. It is the process of an individual or a family obtaining information and advice about a genetic condition that may affect the individual, the individual's progeny, other relatives or the family as a whole, which they can then use to take appropriate, informed decisions about marriage, reproduction, abortion and health management. Genetic counselors should seek to provide facts on genetic information for their clients, putting clients' social habits and religion in consideration before giving advice and without imposing their own views.

As alternatives to marriage prohibition, couples whose tests do prove positive for genetic diseases, could be counseled to choose a number of alternatives, should they still wish to marry. Such alternatives might include avoidance of pregnancy by contraception or sterilization; adoption; donation of a sperm, ovum or preembryo, or motherhood surrogacy; preimplantation diagnosis; diagnosis during pregnancy, for example, chorionic villus sampling (CVS), and amniocentesis, blood testing of both the expectant mother and fetus, and ultrasonography.[4]

A successful example of genetic counseling is the Thalassaemia Control Program implemented in Cyprus, which has succeeded in reducing the incidence of b-thalassaemia major in the country through measures such as health education, carrier screening, premarital counseling and prenatal diagnosis. This success has encouraged other countries to adopt the practice of premarital counseling. Countries or communities practicing such counseling, either voluntarily or by law, are Italy and Greece for thalassaemia, the Ashkanazi Jews for Tay-Sachs disease and some European communities for cystic fibrosis.[4]

Because of the demographic factors and population structure in Middle Eastern countries, e.g. advanced paternal and maternal ages and the high frequency of consanguineous marriages, there is a considerable need for genetic services in order to avoid misinformation and mismanagement of consanguinity on genetic grounds. Premarital counseling is one of the important measures that can help reduce the incidence of genetic diseases in such circumstances.[2,3]

In Saudi Arabia, extensive investigations conducted over several years in different provinces have revealed the wide distribution of the Hb S, a- and b-thalassaemia genes. Studies have shown the high rate of occurrence of these genes in the eastern and western provinces, particularly in the south-west. These areas have a history of malaria endemicity, and despite the fact that malaria has more or less been eliminated; the frequency of the gene's occurrence has remained high.5

Legislation of the Saudi premarital screening program began by a decision of the Saudi cabinet to start the premarital screening program on 21/2/2004 H for all Saudi couples willing to get married. It consists of a compulsory blood analysis for all Saudi individuals willing to marry in order to detect individuals affected by, or are carriers of certain hereditary diseases and to provide them with the appropriate genetic counseling. The screening result is mandatory to complete the marriage license. However, the couple have the choice to proceed for marriage regardless of the result.[6]

The main objectives of this screening program are to decrease the incidence of the common hereditary blood diseases among the Saudi population, to reduce the financial burden and workload due to treatment of the affected patients, and to avoid the social and psychological problems from having children with hereditary blood diseases in families.

As a start, the Saudi premarital screening program includes sickle cell anemia and thalassemia only, which are the most common hereditary blood diseases in Saudi Arabia. In future, other hereditary blood diseases and some sexually transmitted diseases will be added.[6]

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