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CORRESPONDENCE ARTICLE

The impact of genetic diseases on Jordanians: strategies towards prevention

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NATURE OF GENETIC DISORDERS

Genetic disorders are diseases in which genetic factors play an important role in their etiology. They are classified into chromosomal abnormalities, monogenic and multifactorial disorders. While chromosomal abnormalities and monogenic disorders are purely genetic in nature, multifactorial disorders are produced by the interaction between environmental and genetic factors. Although most genetic diseases are individually rare there are many of them. The vast majority are serious, none is curable and relatively few are amenable to satisfactory treatment. A number of surveys have indicated that at least one in every fifty newborns has a major congenital anomaly, one in hundred has a monogenic disorder and one in two hundred has a chromosomal abnormality. However, the prevalence of genetic disorders is quite variable among different ethnic groups and across different age groups.

The control and decline of environmental causes of childhood mortality in western countries threw the genetic causes into greater prominence. Although the picture may be different in third world countries, improvement in living conditions is causing a similar shift. The magnitude of the impact of genetic disorders on all societies is quite significant necessitating their control which can be principally achieved by prevention.

JORDAN AND ITS PEOPLE

The population in Jordan is divided geographically into three distinct sections, all sharing Arabic heritage. The three sections are 1) the urban area of the capital; 2) the Bedouin southern and eastern desert provinces and 3) the agriculturally oriented northern sector. In general, the Jordanian culture, being part of the wider Arabic culture, is dominated by men. The marriage system is mostly consanguineous and polygamy, although accepted, is not widely practiced. The number of children per family is large and the problems of children born out of wedlock and single mothers are close to nonexistent. In rural and Bedouin populations the marriages are usually arranged by parents. Unquestioning obedience to parents by their children, irrespective of their age, is highly valued and is the hallmark of family life across all sectors. Abortion on demand is prohibited and performed only if the pregnancy endangers the mother's life.

Matters of genetic concern have been an integral part of the lives of Jordanians. Several factors contribute to the importance of genetic thinking in Jordan. First, the population is somewhat diverse in its historical ethnicity. Second, the politically oriented massive immigration waves have contributed to the diversity and caused considerable jumps in the census. Third, consanguineous marriages, despite the obvious high risk, constitute over half of the marriages [1, 2]. Fourth, the high birth rate (44 live births/1000 population/year) contributes high absolute numbers of children with genetic disorders and birth defects. Fifth, Jordanians are generally well educated and cultured to the extent of understanding the influence of genetics on their lives. Lastly, despite the remarkable improvement in national health care services including child care, little has been applied directly to genetic disorders and birth defects which contribute significantly to mortality and morbidity both physically and mentally.

One of the outstanding features of the social relationships in Jordan is the existence of consanguineous marriages with considerable frequency [1, 2]. A consanguineous marriage is defined as marriage between individuals who are second cousins or more closely related. However, it is often possible to document lesser degrees of consanguinity quite relevant to pregnancy outcomes, particularly in highly inbred families. The frequency of consanguineous marriages ranges from 50% to 66% in different parts of Jordan [1, 2]. First cousin marriage constitutes about one third of all marriages [1, 2]. Religion, culture, tradition, education, and major historic events affect the frequency of consanguineous marriages but the roles of tradition and historic events seem to dominate in the Jordanian culture. The frequency of consanguineous marriages correlates with an increase in recessively transmitted diseases, congenital malformations and infant mortality. First cousin marriage in inbred families carries an even higher risk for autosomal recessive genetic diseases than first cousin

marriage in noninbred families.

There are very few population-based epidemiological studies that touch on the incidence of genetic disorders amongst the Jordanians. However, it is apparent that similar to the other Arabs the Jordanians have increased frequency of congenital malformations and autosomal recessive disorders [3]. Fortunately, Jordanians are generally strong spiritual believers, believing that the occurrence of disease is God's will. This provides tolerance to genetic diseases and alleviates the feeling of guilt. In addition, faith helps parents overcome their feeling of helplessness thus caring for their affected child with remarkable serenity.

STRATEGIES FOR THE PREVENTION OF GENETIC DISEASES

Genetic counseling is currently the most effective means for prevention of genetic diseases. The main requirement for an effective genetic counseling program is the comprehensive ascertainment of those individuals who are at risk of having affected children so that they can be offered appropriate genetic advice. Although population screening is the obvious method, it is associated with many practical and ethical problems. Routine and accurate diagnosis of genetic disorders is the alternative means for the ascertainment of high risk individuals [4–7]. The families of such individuals can then be informed, screened, and appropriately counseled.

When the family history reveals the presence of an autosomal recessive condition, the question of whether to test individuals for heterozygosity should be considered. For families with a rare disorder a specific test should be designed based either on linked DNA polymorphic markers or on a detected specific mutation. This entails that the gene locus should be known, or the specific mutation identified [8–11]. This offers family members the opportunity of carrier identification and premarital counseling. For common recessive disorders, such as β-thalassemia, familial Mediterranean fever and cystic fibrosis, population-based carrier identification followed by counseling are cost-effective approaches. There is no doubt that personal, familial and even general stigmatization of an identified carrier exists for any screening program. Maintenance of confidentiality, rigorous protection of individual's privacy and informed consent are some of approaches used to overcome such ethical, legal and social issues. With the appropriate technical, medical, and communicative expertise, carrier identification can reduce the burden of untreatable hereditary disorders on the society.

Although termination of pregnancy is prohibited by religion and law, prenatal diagnosis can be a tool for comforting parents, particularly mothers, at risk of having a child with a genetic disease. Prenatal diagnosis, in Jordan is not widely practiced due to the notion that coins it with abortion. Increased public awareness about the benefits of this tool is a necessary strategy towards a wider application.

There is an urgent need to design and apply a model for the prevention of genetic diseases in order to minimize the hazards of these problems amongst the Jordanians. The model should be multiaxial focusing on the following aspects:

(1) Public education on genetic diseases and the factors contributing to its increased frequency. This educational program should emphasize the effect of consanguinity and also deal with applicable preventive measures.

(2) Genetic screening and testing programs for common disorders such as familial Mediterranean fever, hemoglobinopathies, cystic fibrosis, and some inborn errors of metabolism.

(3) Premarital and preconceptional testing and counseling for common disorders. This should also apply to rare disorders that are present in high risk inbred families.

(4) Newborn screening for the prevalent and treatable inborn errors of metabolism, coupled with appropriate early management.

(5) Periconceptional dietetic counseling to minimize the incidence of neural tube defects and maybe others.

(6) The promotion of scientific research efforts for the development of the tools for genetic testing for disorders that are more prevalent in Jordan. This can be achieved through increasing the funds allocated for research activities in the field of human medical genetics. Although this model is partly applied in Jordan, the maximum and impressive benefits will be only reaped with its full application. The model serves all Arabic countries, as well as, other third world countries which share similar cultural and social conditions.

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