

Medical Ethics: Dealing with Families and Communities Affected by Genetic Disease

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Abstract

The Omani population has an astoundingly high number of neonates born with congenital anomalies, deformations, and chromosomal abnormalities. Recent advancements in health care have ensured that the number of affected children who survive infancy has increased significantly. However, most genetic diseases do not have a cure. This makes a significant impact on affected families and includes economic as well as ethical, psychological, and social issues. Families tend to be stressed, and may even be depressed, face financial difficulties necessitated by the increasing medical expenditures, carry a sense of inadequacy and misfortune, and face social disadvantages and stigmatization, forcing a number of families to hide their affected members. Programs targeting the efficient prevention and management of these genetic diseases, therefore, would require certain adjustments in the health care delivery. High ethical principles need to be maintained and promoted. Genetic counseling can be used very effectively, especially since most parents tend to be extremely receptive to counseling, and readily accept family screening for carrier status. Reassuring carriers is also equally important. Educational efforts need to concentrate on making people aware that a genetic disease is not a form of punishment. Important for this is the development and promotion of a new genetic vocabulary that avoids the use of negative connotations for words. Medical personnel need to be trained to empathize with the patients, and to adopt a sensitive approach while dealing with patients and their families. In rural settings, the privacy of the family also needs to be maintained. Psychological rehabilitation of affected patients and their families is another important issue. Finally, an integrated approach needs to be adopted for the efficient management of genetic disorders, which involves health care institutions, social agencies, and community support groups.



الوقاية من الأمراض الجينية والوراثية من خلال توعية الأسر والمهتجات

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ملخص

يشكل المواليد الجدد المصابين بالتشوهات الخلقية والاضطرابات الوراثية الناجمة عن خلل في تركيب الكروموسومات أعداداً هائلة في المجتمع العماني. ولكن أخذ عدد الوفيات لهؤلاء المواليد بالإنخفاض مؤخراً وذلك بسبب التطور الحادث في الرعاية الصحية. ولكن إلى الآن لا يوجد علاج لمعظم تلك الأمراض الوراثية، وبالطبع يبقى تأثيرها الكبير على العائلات المصابة واضحاً في النواحي الاقتصادية والنفسية والاجتماعية. حيث تشعر هذه العائلات بالضغط النفسي وقد تشعر بالإحباط والاكتئاب لما تواجهه من مشاكل مالية ناجمة عن تكاليف الرعاية الطبية الباهظة. كما قد تحمل هذه العائلات احساساً بسوء الحظ وعدم القدرة على التكيف في المجتمع وذلك لما تواجهه من مشاكل اجتماعية ناجمة عن عدم قبول المجتمع لأطفالها المصابين الذين قد يتصفون بعدم الفائدة وقد يجلبون العار لعائلاتهم. وقد يدفع هذا كله العائلات لعدم اظهار أبنائهم المصابين أمام المجتمع. وبالتالي فالحاجة ملحة لتنظيم برامج تهدف إلى الوقاية والعلاج من هذه الأمراض الوراثية مع مراعاة وضع ضوابط معينة لهذه البرامج. وتساهم الاستشارة الوراثية بشكل فعال في هذا المجال، خاصة أن معظم الآباء متفتحي الذهن للاستشارة الوراثية ويقبلون بفكرة الكشف عن حاملي المرض من أبنائهم الغير مصابين. فالكشف عن حاملي المرض من غير المصابين وطمأننتهم له أهمية تعادل أهمية الكشف عن المصابين. ويجب أن تركز الجهود التثقيفية المبذولة على توعية الناس بمبدأ أن الأمراض الجينية ليست عقاباً. وتأتي أهمية هذا المبدأ من منطلق التشجيع على إيجاد مصطلحات جديدة بعيدة عن السلبية عند الحديث عن الأمراض الجينية. ويتطلب ذلك تدريب الطاقم الطبي على تبني منهج حساس في كيفية التعامل مع المرضى وعائلاتهم. كما يجب الحفاظ على خصوصية العائلة خاصة في المناطق الريفية. كما تشكل قضية إعادة تأهيل المرضى وذويهم من الناحية النفسية أهمية بالغة. ولكي تتجح هذه الخطى، يتطلب إدماج مثل هذه البرامج تبنيها من قبل مراكز الرعاية الصحية والهيئات الاجتماعية وجمعيات المجتمع.



Introduction

In the past thirty years, GCC countries have witnessed remarkable social and economic growth which is best reflected in the well-organized and efficient health care system. With these achievements, countries registered an epidemiologic shift in disease pattern. There has been a significant decrease in the incidence of communicable diseases and in the mortality and morbidity rates of infants and children under 5 years. The national incidence of stillbirths and of congenital anomalies in Oman was recorded as 0.9% and 16.9% respectively (Alwan and Modell, 1997; Modell, 2003; Rajab *et al.*, 2005). The figures are astoundingly high when the incidence of congenital malformations, deformations and chromosomal abnormalities are considered together (73 / 1000 births). The estimated incidence of children born with congenital and genetic disorders is 7% as compared to 4.4% in Europe. Prevention of mental retardation, hemoglobin disorders and handicapping genetic diseases is becoming a priority, and the projected cost of care in the next decade has an impact on allocation of funds for preventive services. The impact on families affected by genetic disease is significant, but the amount of human suffering related to genetic diseases in families cannot be measured in dollars.

New ethical, medical, social and economic issues are emerging with new genetic technologies. Treatment and prevention should, at all times, be complementary aspects of a holistic service aiming to help families with a genetic problem. The management of individuals affected with genetic diseases goes well beyond a short period of an acute episode of illness, and psychological rehabilitation for the affected and their relatives can be as important as acute treatments, and often, even more important.

Further studies of efficient methods of health education in genetic matters, and the impact of specific words in the Arabic language are required. In order to structure efficient prevention programs, few important adjustments in health care delivery are needed. These should be based on the availability of comprehensive genetic diagnostic facilities, upgrading medical personnel in medical genetics, and the provision of comprehensive rehabilitation facilities for those affected by genetic diseases. The understanding and respect to people's feelings and believes appropriate for the local community health education, ethical approaches, and integrated support to the families affected by genetic illness will pave the road for the success of prevention programs.

Recent Change in Health Care

The presence of a comprehensive health care system and the improvement in quality of life in GCC states has resulted in better survival of children born with disadvantages, and a progressive increase in the prevalence of genetic diseases and handicap in the community. Medical services have reached such a standard that many patients who would formerly have died are now surviving into adult life. Before genetic diagnoses could be made, illness and death of the children was attributed to weak health, failure of breast-feeding, bad luck or the evil eye. In the past, the scale of the problem of congenital/genetic disorders (including hemoglobin disorders) was hidden in the high infant mortality, because most affected infants died without being diagnosed. Now, the majority are diagnosed and provided with the best possible treatment. As a result, to the extent that treatment ensures survival, the number of surviving affected children increases by the number born annually. The cumulative number of patients requiring care is therefore rising rapidly.

Children are important in traditional Muslim communities being considered an important part of the prosperity and pride of the family, as well as the beauty of life. Childbearing remain of prime importance for the majority of women and most women are totally devoted to bringing up children and caring for the family.

The concept of "genetic disease" is fairly new in the Islamic world and Clinical Genetic services are only recently becoming available. Not long ago illness and death from genetic cause was unknown. In the absence of accurate genetic diagnosis the terms, "dysmorphic" or "cerebral palsy" are still often used to explain childhood illness, and the death of children with undiagnosed genetic diseases was attributed to other causes such as "infection" or "cardiac/respiratory failure. Families with genetic disease need to learn about the correct causes (and, occasionally) of possible treatments for their affected children. For the most of genetic diseases no radical cure is available, which cause additional psychological and social problems.

It is an understandable clinical experience that whereas a mother with one affected child would be stressed, a mother with two or three affected children may become psychologically disturbed or depressed. Overstressed and tired mothers who care for their handicapped children consequently are unable to give enough attention to other children, who may feel neglected. Fathers may tend to blame



themselves or their wife for producing the abnormal children, can be very angry, and may consider another marriage. In the end, the whole family suffers from the daily strain of observing and caring for children who are handicapped or suffer from an incurable illness.

Disillusioned parents often wear themselves out visiting many hospitals searching for someone who can offer treatment or explain to them how to deal with the child. In addition, financial difficulties often arise, especially in families with multiple affected children. Disproportionately expensive treatments (e.g., new medications and incontinence pads) are additional costs. Extra help is required at home with feeding, washing, and coping with abnormal behavior. Children with genetic disorders fall ill frequently and require regular hospital visits. A father with multiple affected children may be absent from work (to keep numerous appointments) and can sometimes lose his job because of these frequent absences.

Families with genetic diseases also have to deal with social disadvantages, including the refusal of marriage with family members. This affects primarily females and can have a profound effect on their life.

Families tend to suffer from stigmatization. Disadvantaged persons are seen to negatively deviate from socially constructed norms and expectations and may evoke negative responses and courtesy from individuals/groups of people such as intrusive inquiries, staring, pointing, comments and devaluing remarks. The family may suffer social withdrawal of family and friends.

The modern term "genetic" or "*wirathi*" seems to carry negative connotations in the Arabic language and this is reinforced by the confusion and uncertainty about why the disease appeared and why modern medical care is unable to correct it. Lay peoples' responses to genetic disease are associated with feelings of fear, misfortune, shame, guilt, anger, isolation and blame. These feelings are often accentuated by negative connotations in the mass media and among medical professionals.

Knowing the adverse aspects which come with stigmatization and publicity, parents tend to hide the real problem and deny genetic disease in their own family. They sometimes try to hide handicapped children within the household from other family members and medical professionals. Parents may not accept a genetic diagnosis and may not wish to discuss it.

A special situation exists in rural communities that are relatively small and consist of large extended families. There, parents of affected children feel inadequate, having failed to produce sufficient normal children. The diagnosis of genetic disease appeals to them as a misfortune that will influence their entire lives and they may wish to keep it secret. Once it is known, they have difficulty in facing or confronting their community (where everybody knows everybody else and news is spread around in a moment).

Most young couples are open to information and counseling and easily understand the concept of recessive inheritance. Parents are eager to learn about ways to avoid disease and some readily accept family screening for carrier status in order to avoid future marriages between carriers within the family. Using knowledge for prevention is an efficient tool.

Proposed Solutions

Promotion of Ethical Principles: Maintaining High Ethical Principles include practicing the highest standards of confidentiality, and respecting the autonomy of the individual couple, and their right to have complete and accurate information. Additional measures are avoidance of negative terminology, minimizing the chance of psychological harm and anti-stigma counseling. The core principles of ethical genetic counseling, excluding directive or coercive advice, should be observed and the issue of prevention should come as secondary to the stated wishes of the patient or couple.

Genetic Counseling: Genetic Counseling has the potential to be very useful in reducing parental fears of an obscure and frightening force behind genetic illness (Raeburn, 1998; Modell and Darr, 2002; Modell *et al.*, 2003). Once they are given the true biological explanation (e.g., of how DNA changes can alter body proteins), they can put the illness in their family into some sort of context. Most couples with affected children are open to information-giving and counseling, and easily understand the concept of recessive inheritance. Parents are then eager to learn about ways to avoid disease; for example some then readily accept family screening for carrier status in order to avoid future marriages between carriers within the family (Al-Odaib *et al.*, 2003; El-Hazmi, 2004a). In the setting of premarital clinics positive attitude should be adopted. Special attention



and support should be given to women, especially those who are carriers. Women readily accept the responsibility for the health of their future family and therefore women must be fully informed about possible prevention along with their husbands, so that decisions about any future methods of prevention are shared. It is necessary that clear explanations be given about possibility of predictive genetic tests.

Reassurance for carriers is important. The explanation should be given that to be a carrier is not an illness and not a God's punishment. Being a carrier is not shameful and is often associated with advantage for survival. In the case of sickle cell disease, carrier is selected by nature for survival against malaria. A carrier will not develop illness later on. The advantage of knowing one is a carrier is to be able to plan a healthy family and avoid genetic disease.

Health Education: Using Genetic Knowledge for Prevention

Educational efforts may wish to increase understanding by communities that genes can have deleterious effects and cause diseases throughout the life span. Above all, people throughout society must be helped to understand that everyone carries a number of "variant" genes, which may induce severe illness in our children, or may show effect in later life. It is important to stress that a child with genetic disease is not a punishment from God; the gene for a recessive disease is inherited from both parents but parents are not responsible for the combination of genes they have and can not be blamed.

New methods of health education should be developed and promoted. A new genetic Arabic vocabulary may need to be established, which is less damaging to patients and their families. In particular, much understanding of the impact of specific words in the Arabic language is required.

A paradigm shift in medical education is needed, one with more emphasis on training future physicians to enhance their empathy skills and to learn to view patients as persons, not just cases.

Sensitive Approaches

Families with handicapped children should not be blamed, criticized, judged or ridiculed, but should be supported in all ways possible. Health practitioners need to adopt a sensitive approach in providing health information paying attention to their vocabulary to ensure that unintended meanings are not communicated. The management of those affected with genetic diseases goes well beyond a short period of an acute episode of illness. Psychological rehabilitation for the affected and his relatives can be equally important as acute treatments, and often, even more important.

Every effort should be made to minimize diagnostic uncertainty (WHO, 2004; Condit *et al.*, 2004). The value of genetic diagnosis encompasses (beyond clear plan of treatment) family planning, recurrence risk for siblings, and a significant emotional relief for the parents. Each family has the right to an explanation for a handicap, to appropriate investigation and to information as to how molecular diagnosis may open the way to prevention (Al-Odaib *et al.*, 2003; El-Hazmi, 2004a). Parents need to be educated about how genetic disease recurrence could be prevented, and how relatives can also avoid the same condition.

Special consideration must be given to preserving the privacy and integrity of families living in rural communities. Unintended "advertisement" of their genetic disadvantage may result if professionals visit and pay attention excessively to families with genetic disease with offers of counselling, premarital tests or research (El-Hazmi, 2004b).

It would be essential to discuss with parents of newly diagnosed children with genetic disorders how they wish to maintain confidentiality as well as what support services they need. Families need assistance to develop coping strategies with follow-up.

Integrated Management

An integrated approach to the management of the children with disadvantages, requiring support from primary health care institutions, social agencies and community support groups, is an essential adjunct to genetic counseling services. The community support groups and associations are able to play a major role in helping parents to manage disabled children in terms of sharing practical experience with other parents about therapies and increasing relevant skills, and with respect to emotional aspects such



as getting a sense of belonging to a community.

Only if it is accompanied by visible commitment to the care of affected people will a prevention programme be credible and able to gain the confidence and co-operation of affected families (Rajab and El-Hazmi, 2007).

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