Strategies for low and medium resource countries in the community care and prevention of birth defects:

Jordan model

A proposal for a national strategy on the care and prevention of birth defects in Jordan

Prepared by: Hanan Hamamy[1], Sana Al-Hait[2], Ala'din Alwan[3]

Based on the workshop: **"National strategies for the prevention of genetic and congenital disorders"** held by the Ministry of Health in collaboration with the World Health Organization on the 2nd of October 2005

[1] National center for Diabetes, Endocrinology and Genetics, Amman, Jordan

[2] Section for Prevention of Genetic and Congenital Disorders, Directorate for Disease Control and Prevention, Primary Health, Ministry of Health, Amman, Jordan

[3] World Health Organization, Amman, Jordan

How to cite this article:

Hamamy H, Al-Hait S, Alwan A. Strategies for low and medium resource countries in the community care and prevention of birth defects: Jordan model. A proposal for a national strategy on the care and prevention of birth defects in Jordan. Geneva Foundation for Medical Education and Research [Internet]. 2009 Aug 27 [cited 2009 Aug 27]. Available from: http://www.gfmer.ch/Medical_genetics_education_research/Strategies_care_prevention_birth_defects_Jordan_model_Hamamy_2009.pdf

Summary

Jordan, like other Arab countries, is faced with major challenges in providing comprehensive and up-to-date health services in a rapidly advancing field such as genetics. Contrary to some widely held misconceptions, scientific evidence and experience in countries indicate that there are effective evidence-based strategies for the care and prevention of common genetic and congenital disorders at the community level which depend on a combination of basic public health measures, and the education and involvement of the primary health care network. Primary health care services in Jordan are delivered through an extensive primary health care network which could facilitate the integration of community genetics approaches within the existing health system.

Strategies for the Care and Prevention of Genetic and Congenital disorders

- Full commitment of policymakers and provision of adequate managerial support.
- Integration of community genetic services into primary health care through structured education and training of primary health workers and updating the nursing and medical college curricula on human genetics and community genetics.
- Strengthening human resources by promoting more specialists in the field of genetics such as clinical geneticists, genetic counsellors, social workers and laboratory personnel.
- Education of the public through scientific evidence-based messages.
- Defining the ethical, legal, religious and cultural issues in formulating genetic services that conform with the needs and beliefs of the population, and formation of special committees to formulate guidelines for screening programs and for selective pregnancy termination of affected fetus.
- Initiation and monitoring of population screening programs such as newborn screening for phenylketonuria and congenital hypothyroidism, premarital screening for thalassemia carriers, prenatal screening for Down syndrome and neural tube defects.
- Initiation and monitoring of a national birth defects registry.
- Introducing new technology and strengthening of existing genetic service facilities.
- Provision of proper management and care for the affected.
- Provision of sufficient, good quality and affordable special schools for the disabled.

The terms 'birth defects' and 'genetic and congenital disorders' are used interchangeably in this document

Introduction

Genetic disorders and congenital abnormalities constitute a major health problem worldwide. They occur in about 2%-5% of all live births, account for up to 30% of pediatric hospital admissions and cause about 50% of childhood deaths in high resource countries.

Contrary to the generally held belief, the limited epidemiological data presently available from Jordan and other Arab countries indicate that congenital and genetically determined disorders may occur in frequencies similar to or higher than those reported in high resource regions of the world (1,2).

The magnitude of the problem of genetic and congenital disorders in Jordan can be attributed to several factors including:

• The high consanguinity rate in Jordan, which increases the risks of recessively inherited diseases;

• The high frequency of haemoglobinopathies and glucose-6-phosphate dehydrogenase deficiency, possibly related to selective advantage of carriers against falciparum malaria and other as yet unknown factors;

• The social trend of continuing to bear children up to menopause, which increases the predisposition to chromosomal trisomies such as Down syndrome, owing to advanced maternal age; also, the incidence of certain autosomal dominant disorders increases with advanced paternal age;

• The large family size in Jordan may contribute to the increase in the number of affected children in families with autosomal recessive conditions;

• The general lack of public health measures directed at the prevention of congenital and genetically determined disorders and the dearth of genetic services and inadequate health care prior to and during pregnancy.

Current impediments that limit the availability of genetic services in Jordan

Jordan, like other Arab countries, is faced with major challenges in providing comprehensive and up-to-date health services in a rapidly advancing field such as genetics. These impediments include:

- Paucity of resources.
- Presence of other competing priorities, such as increasing needs for the control of communicable diseases and non-communicable chronic disorders as cardiovascular diseases, cancer, and diabetes.
- Insufficient number of trained health professionals in the area of medical genetics.
- Inadequate data on the real magnitude of genetic and congenital disorders , and their health and economic burden.

- Misconceptions that the control of common genetic disorders is too expensive and always linked with sophisticated high technology, limiting its introduction to the general public
- Low genetic literacy among the health sector and the public with lack of awareness about genetic risks and possibilities for care and prevention of birth defects.
- Community services may be restricted by certain cultural, legal and religious limitations such as the cultural fear of families with genetic diseases to be stigmatized within their community and the legal and religious restrictions on selective abortion of an affected fetus.

A comprehensive national initiative on the development of basic genetic services is particularly needed in Jordan because of the following:

- *Declining morbidity and mortality from infectious diseases, and declining infant mortality in Jordan.*
- *Good coverage of reproductive primary health care programs in Jordan*.
- *The need for better understanding of the relationship between consanguinity and genetic disorders*. Consanguineous marriages are common in Jordan, with 20-30% of all marriages contracted between first cousins (3,4).
- *The high rate of hemoglobinopathies and other single gene disorders* in Jordan and the great potential of establishing cost-effective care and prevention programmes. It is estimated that among males, 12% may be affected by glucose-6-phosphate dehydrogenase deficiency, and that the carrier rates for beta thalassemia, alpha thalassemia and sickle cell anemia are about 3-6%, 1-2% and 1-4% respectively (5-9).
- *The public is becoming more aware of effective services like genetic testing and risk assessment and is now demanding the introduction of such genetic services*. Many families seen at the genetic clinics request better access to effective facilities for diagnosis, care and for prevention.
- *Genetics is becoming an essential part of most medical specialties.* Increased awareness about the role of genetics in disease and the great advances made in medical genetics in recent years have had a considerable impact on the practice of medical genetics.

Strategies for the care and Prevention of birth defects in Jordan

Contrary to some widely held misconceptions, scientific evidence and experience in countries indicate that there are effective evidence-based strategies for the care and prevention of common genetic and congenital disorders at the community level. Many of the preventive interventions do not require sophisticated and expensive technology and can be integrated into the existing health system in Jordan. While the overall objective of a national programme is the prevention and care of genetic and congenital disorders in the community, the strategies adopted to achieve this objective should be carefully selected to match the unique demographic, cultural and religious characteristics of the population, and take into consideration the priorities set, and the resources available.
Prevention of congenital and genetic disorders at the population level depends on a combination of basic public health measures, and the education and involvement of the primary health care network. Such measures include interventions that target the whole

population with the public health goal of reducing the burden imposed by genetic and congenital disorders. They basically focus on health promotion and preventive programs implemented at the primary health care and community levels and supported by an affordable and cost-effective package of secondary and tertiary care services.

Generally, prevention of genetic and congenital disorders can be addressed at three levels:
Primary prevention: premarital screening and counselling and preconception counselling
Secondary prevention: prenatal counselling, screening, and testing with the option of termination of affected fetus or prenatal and neonatal management.
Tertiary prevention: Newborn screening with proper management can be considered as secondary or tertiary prevention. Care of the affected and prevention of complications and rehabilitation of the handicapped can be done at the primary health care or at higher levels.

The Strategies for the care and Prevention of birth defects comprise:

1- Commitment of policy makers

Political will and commitment is needed for funding, planning and managing the care and prevention programmes. Policymakers become more aware of the health burden of genetic and congenital diseases when reliable data on magnitude and characteristics become available. Epidemiological studies on frequency of the disorders and their impact on public health and social life should therefore be conducted without delay. Decisions should be made on whether genetic services should be comprehensive or start with specific priorities dictated by local needs and available resources. Prevention programmes planned, besides being cost effective, should take into consideration local beliefs and social attitudes. For example, premarital screening and preconception counselling as preventive measures are more acceptable than therapeutic abortion in Jordan.

Development of a situation analysis report based on currently available data would help in focusing on priorities in the prevention program. **This report includes:**

- Data on the frequencies of genetic and congenital disorders.
- Data on available genetic services (clinical and lab).
- Data on number and capacities of institutions for handicapped children.
- Data on the available Medical and Nursing college curriculum on genetics and genetic services.
- Data on available human resources in clinical and laboratory settings in genetics.

2- Integration of community genetic services into the primary health care system

Integration of community genetic services into existing health care systems can be feasibly implemented in Jordan. Establishing vertical programs is difficult with a high demand for sustainable funds and human resources. Integration of public health approaches into the existing primary care and reproductive health clinics is probably the most appropriate, sustainable and cost-effective approach. Although some additional training and resources will be required, the potential benefits are considerable. An example of the integration of

community genetic services into primary health care programs is the inclusion of preconception counseling and screening in reproductive health clinics. The approach includes preconception information (nutrition, maternal infections, Rh status, parental age, maternal disease, teratogenic drugs and chemicals,), referral of couples at high risk to specialized centers, carrier screening, and newborn screening programs (10). Community genetic services could also concentrate on providing family oriented services such as genetic counseling. The implication is that genetic counseling has a particular potential for providing help to families with genetic diseases due to the high rate of consanguinity and the large family size in Jordan.

3- Strengthening human resources: Education and training in the context of community genetic services

Strengthening human resources through training and education of the health sector will provide the main area of work in implementing community genetic programs.

Review and updating of the medical and nursing college curricula related to the practice of human genetics would on the long run supply the manpower required for the prevention programs. More emphasis could be placed on practical guidelines of how to approach genetic and congenital disorders at the primary health care level. Priorities in teaching could be given to diseases common in the country. Strengthening the continuing education programs and their dissemination throughout the country would provide essential information and training for all health personnel. There is also the need for special training courses for laboratory skills development.

Primary health care workers are not adequately trained to deal with common genetic disorders. They need to acquire the necessary skills in first level counselling, so that they can be involved in providing premarital and preconception counselling, and counselling prior to newborn and carrier screening. Genetic counselling for individuals or families at high risk or those with unfavorable test results is given provided by specialists in the field.

Health workers need to be educated in basic scientific principles of genetics and in the ethics and practice of genetic counseling. The World Health Organization Eastern Mediterranean Regional Office publication Community control of genetic and congenital disorders (1) provides an outline of the basic requirements to strengthen the capabilities of primary health care workers in preventing genetic diseases. These include:

- Training in taking and recording a basic genetic family history, taking account of the complexities of large families with multiple consanguineous marriages;
- Guidelines on detecting possible genetic risks (e.g. history of previous stillbirth, neonatal death, congenital malformation, multiple abortion or hereditary blood disorder in the family);
- Guidelines on lines of referral and clear information on specialist services available;
- Training in the basic ethical principles and techniques of genetic counseling.
- Guidelines in counseling on consanguineous marriages Evidence-based guidelines regarding consanguinity in genetic counseling settings should be developed and educational material to clarify these guidelines should be prepared. Consanguinity is a sensitive issue in communities where half of all marriages are consanguineous, and the social benefits of consanguinity may be seen by some to outweigh the adverse health effects.

4- Education of the public

Education of the public is a definite priority since ignorance and misconceptions could be barriers to the implementation of community genetic programs. Organized information, education and communication need to be addressed to the population in general through different channels that include school curricula and media messages. Education is a prerequisite to screening programs, since a well-informed individual can take responsible decisions.

1- **School curricula** could include:
Basic principles of Human genetics
Information on common genetic disorders in the community and on screening programs

2- **Population Education** could be addressed through:
Information acquired through trained primary health care workers. Premarital, prescreening and preconception counseling could be the best educational tool if made accessible to all those in need.
Health pamphlets addressing methods for the prevention of genetic and congenital disorders
Mass media educational campaigns must be scientifically based and appropriately delivered. These campaigns must respect local cultural and religious beliefs and avoid controversial issues and convincing messages. Inaccurate messages could do more harm than good and would eventually interfere with the successful implementation of the program.

5- Ethical, legal, religious and cultural issues

Defining ethical standards must be a component of any community genetics programme. In medical genetics, progress and new discoveries are running ahead of ethical formulations. The public in general, religious leaders, many politicians and some health professionals do not feel comfortable with the very rapid advance in the science of genetics and molecular biology It is important to alleviate fears, so that community genetic services become widely acceptable and sought.

Generally the main ethical standards that need to be addressed are:

- Genetic services should reach all those in need
- Education of the public
- Services should not contradict cultural and religious beliefs
- Definition of guidelines for prenatal genetic diagnosis
- Definition of guidelines for selective therapeutic abortion of an affected fetus
- Definition of guidelines for premarital screening programs
- Definition of guidelines for newborn screening programs
- Definition of guidelines for premarital counseling
- Definition of guidelines for counseling and media messages on consanguinity

6- Population screening programs

These could include:

- Newborn screening for hypothyroidism, phenylketonuria, G6PD deficiency,other metabolic disorders
- Premarital screening for beta thalassemia and other common autosomal recessive disorders
- Antenatal screening for Down syndrome, neural tube defects and major malformations.

7- Birth defects Registry

There are certain requirements that need to be addressed regarding the initiation of Genetic Registries, such as:

- What conditions should be initially registered
- Training personnel
- Defining responsibilities
- Recruiting experts for planning and implementation of the project
- Training physicians and nurses on examination of newborns to detect congenital malformations

8- Genetics Centers and introduction of new technology

The establishment of a national centre for the control of genetic and congenital disorders aims at introducing new technology that shapes the diagnostic and therapeutic facilities of present day genetics. The national genetic center is also responsible for the dissemination of information and for advising on community genetic services. Research in the field of genetics in each country is essential to define local issues, to indicate priorities and to solve emerging problems. There is a need to study the already existing experiences regarding genetic services in the area and the reaction of the public to these services. Genetic testing is currently a major issue in preventive medicine; the possibilities for including genetic testing in community genetic programs in Jordan could be investigated.
The molecular revolution that characterized the last two decades has introduced into medical practice many procedures that aid in the diagnosis, care and prevention of genetic diseases and it is important for low and medium resource countries to take account of these developments. .

9- Provision of proper management and care for the affected at all levels of health care including primary health care

10- Provision of sufficient, good quality and affordable special schools for the disabled

The overall goal of the strategy would be to monitor yearly the frequencies of affected births of genetic and congenital disorders and to achieve a progressive decline in the national rates of birth defects, with provision of the best available care for the affected.

References

1. Alwan A . & Modell B. Community Control of Genetic and Congenital Disorders, EMRO Technical Publications 24, World Health Organization, 1997
2. Teebi A and Farag T. (eds.) Genetic Disorders Among Arab Populations, Oxford Monographs on Medical Genetics No.30. NY: Oxford University Press, 1997.
3. Khoury SA, Massad D : Consanguineous marriage in Jordan. Am J Med Genet 1992 ;43:769-75
4. Hamamy H, Jamhawi L, Al-darawsheh J, Ajlouni K : Consanguineous marriages in Jordan: why is the rate changing with time? Clin Genet. 2005 ;67:511-6.
5. Babiker MM, Bashir N, Sarsour N : Prevalence of thalassaemia in schoolchildren in north-eastern Badia, Jordan. East Mediterr Health J 1999; 5:1165-70.
6. Talafih K, Hunaiti AA, Gharaibeh N, Gharaibeh M, Jaradat S : The prevalence of hemoglobin S and glucose-6-phosphate dehydrogenase deficiency in Jordanian newborn. J Obstet Gynaecol Res 1996; 22:417-20
7. Sunna EI, Gharaibeh NS, Knapp DD, Bashir NA: Prevalence of hemoglobin S and beta-thalassemia in northern Jordan. J Obstet Gynaecol Res 1996:22:17-20.
8. Bashir N, Barkawi M, Sharif L, Momani A, Gharaibeh N : Prevalence of hemoglobinopathies in North Jordan. Trop Geogr Med 1992 ;44:122-5.
9. Bashir N, Barkawi M, Sharif L : Prevalence of haemoglobinopathies in school children in Jordan Valley. Ann Trop Paediatr 1991;11:373-6
10. Hamamy H & Alwan A. Genetic disorders and congenital abnormalities: strategies for reducing the burden in the Region, Eastern Mediterranean Health Journal, 1997, 3(1): 123-132.