Report

Early recognition and intervention for prevention of disability and its complications

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Introduction

Birth defects, i.e. congenital disorders present at birth, occur in 2%–4% of all neonates [*1*]. These abnormalities may be caused by environmental factors that affect the mother, or are inherited via abnormal genes from the carrier or affected parent. Such disorders account for about 20% of deaths during the neonatal period and a higher percentage of morbidity in infancy and childhood [2]. With the dramatic decrease in infant mortality due to improvement in the control of infections and malnutrition-related disorders, chronic disabling conditions are an emerging challenge facing developing and industrialized nations.

Occurrence and prevalence

Prevalence rates of the various types of disability vary in different populations. Although only a limited number of national surveys have been conducted to identify the etiological factors in the development of disability [*3*], it is generally believed that 76% of disability is caused by genetic factors [*4*]. In one survey, it was shown that 21.9% of cases of disability resulted from prenatal damage; 3.0% arose from perinatal factors; 29.0% were acquired during infancy and early childhood and 47.0% had no known cause [*5*]. The overall prevalence rate of disabled children was 2.7% and age-specific prevalence rates showed an increase with age [*5*].

Etiological factors

Disability may be developmental or acquired and may arise from prenatal damage, perinatal factors, acquired neonatal factors and early childhood factors. These may include genetic factors, infections, traumatic or toxic exposure or nutritional factors which result in perinatal or postnatal damage.

Etiological factors of disability among children at different stages are:

- Prenatal: genetic factors, genetic diseases, developmental malformation, maternal age, maternal diseases, drugs/medicines/chemicals/radiation, consanguinity, ethnic group.
- Perinatal: low birth weight/prematurity, obstetric complications, trauma during labour, asphyxia, intracranial haemorrhage.
- Postnatal: infections (poliomyelitis, tuberculosis, meningitis, encephalitis),

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Received: 03/06/96; accepted: 23/10/96

endemic diseases (goitre, cretinism), accidents, malnutrition, poisoning, tumours, environmental factors, psychosocial problems.

The causative factors leading to disability are heterogeneous and complex, and their contribution in producing disability may differ in different populations [*6*]. Furthermore, the etiology of a substantial percentage of disability remains unknown.

Prevention

Every human society requires that its future generation be healthy. This depends on the birth and rearing of healthy children. To this end, preventive screening for genetic disorders, including developmental disabilities, is an essential component in uncovering possible disorders early, thus enabling timely medical intervention.

Such efforts are also required in order to reduce the expression and severity of disability. The ability of a physically or mentally disabled child to cope with and adapt to everyday life may be minimal compared to that of a normal child, and the disabled child may continuously suffer from trying to perform the functions so normal to others. This can have a major influence on the personality of the child which in turn can affect normal growth and development. Of particular distress is the fact that some disabled children never reach adulthood and some are at risk of developing other associated complications which may further disrupt their social and emotional development. These children and their families are under continuous mental and physical stress and require comprehensive services in order to help the children have a near normal life [*7–11*].

In addition, many disabled people require a continuous health care system, home help and other supportive services, which makes care programmes very costly. The management of childhood disabilities requires substantial medical, educational, social and rehabilitative care [*6,7,12*]. The cost of preventive efforts is substantially lower and thus cost–effectiveness favours the prevention approach.

Stages of prevention

Endeavours for controlling disability can be categorized as primary, secondary and tertiary prevention.

Primary prevention

This involves the prevention of the manifestation of the disability. It may be universal (i.e. prevention desirable for everyone), or be restricted to a selected population (i.e prevention recommended for high-risk groups) or to an indicated population (i.e. prevention in individuals with an identified risk).

Primary efforts are directed toward reducing the actual occurrence of disabilities and they employ measures that prevent the conception of a disabled individual or delay the disabling process. Primary prevention efforts include:

- genetic counselling
- immunization programmes
- improved prenatal, perinatal and postnatal care
- regulations and legislation
- other related means.

Genetic counselling

Genetic counselling is an essential part of primary prevention strategies. It is the process of providing information on genetic (recurrence) risk, the nature and consequence of genetic disorders and the means

available for the prevention of transmission of defective genes [*13*]. Within this framework, there are three major aspects essential to effective counselling [*14–18*] which are:

- diagnostic aspects, where an accurate diagnosis is required for a secure foundation for advice
- estimation of risk
- preventive or ameliorative measures to ensure that those who are advised will benefit.

One of the prime requirements of an effective genetic counselling programme is to ascertain which individuals are at risk of having an affected child so that they can be offered advice. Genetic screening and counselling prior to conception is important for the control of genetically determined disabilities [*16*]. Many studies have shown that genetic counselling of carriers, premarital couples, couples with a disabled child and other family members produces better understanding of the issues involved and hence has an impact on the subsequent reproductive decision. Several studies have shown that the number of children born with a serious genetic disorder leading to disability decreases significantly following genetic counselling.

The best example of the successful application of this prevention strategy to prevent the birth of homozygotes for a certain disorder is the thalassaemia control programme in Cyprus. Through health education, together with population screening and genetic counselling of carriers and prevention of carrier marriages, it was possible to reduce the homozygous affected births from 53 per 8594 births in 1974 to 0 per 10 752 in 1988 and 2 per 10 830 in 1990. The percentage of prevention achieved was 1.8% in 1974, 100% in 1988 and 97% in 1990 [*19*]. A similar approach when applied to other disorders could be equally successful.

Immunization programmes

Programmes of general immunization during infancy have led to a remarkable decrease in, or in a few cases a complete absence of, several infectious diseases that used to be a major cause of disability. These include poliomyelitis, tuberculosis, meningitis and encephalitis [*20*].

Improved prenatal, perinatal and postnatal health care

This aspect of prevention concentrates on the management of maternal risk, factors at the time of delivery and support for the premature or compromised neonate.

Regulations and legislation

Certain health regulations and legislation, such as mandating immunization of infants, also play an important role in primary prevention.

Secondary prevention

Secondary prevention strategies aim at reducing the duration or severity of disability. These activities provide early identification of the disabling condition followed by prompt treatment and intervention to minimize the development of disability. These strategies can be applied either at the prenatal or neonatal level. Some of the conditions that can be diagnosed during the prenatal and neonatal stages are listed in Table 1.

Neonatal screening (organized examination of all neonates in order to diagnose specific disorders so that they can be treated) is a well established preventive approach and includes both clinical and biochemical screening. In some countries, such information is available but in others,

there is no information on detection frequency at birth of genetic disorders.

At the neonatal level, screening of neonates and proper intervention in those affected have been successful in reducing disability. Examples of the usefulness of such action are shown in Table 2. The best known example of secondary intervention is that of neonatal screening for phenylketonuria (PKU), other aminoacidurias, hypothyroidism, the thalassaemias and other haemoglobinopathies. In PKU and other aminoacidurias, once the baby is diagnosed as having an abnormality, proper measures are taken by providing special diets.

Biochemical screening was first introduced for PKU in 1966 when it was shown that a low phenylalanine diet started in the first week of life prevents severe mental retardation. To screen for PKU, blood sam-

Table 1 **Tests used in early identification of disability or diseases that may lead to disability**

Stage	Test	Diagnosis
Prenatal	Maternal α-fetoprotein	
	–elevation	Neural tube defect
	–decrease	Trisomy 21, trisomy 18
	Maternal unconjugated estral decrease	Trisomy 21, trisomy 18
	Maternal human chorionic gonadotropin elevation	Down syndrome
	Ultrasonography	Down syndrome and other physical defects
	DNA analysis of amniotic fluid, chorionic villi or fetal blood	Several inborn errors of metabolism
	Cytogenetics	Numeric and structural anomalies of the chromosomes
Neonatal	Estimation of phenylalanine	Phenylketonuria
	Estimation of other amino acids	Aminoaciduria
	Estimation of the level of thyroid hormones	Hypothyroidism
	DNA analysis	Several inborn errors of metabolism Cystic fibrosis Thalassaemias Other haemoglobinopathies Duchenne muscular dystrophy Fragile X syndrome Huntington disease Myotonic dystrophy Haemophilia
Carrier detection	DNA analysis Specific tests for each disorder (enzyme assays, electrophoresis, protein estimations, others)	As above

ples are usually taken by heel prick between 5 and 10 days after birth, when the body's metabolism has stabilized sufficiently for the results to be reliable. Screening for PKU is now established in several countries and screening for other abnormalities is also carried out, particularly since the advent of recombinent DNA technology. Hypothyroidism diagnosed during the neonatal period is treated by hormone replacement therapy, which encourages normal development and prevents complications such as mental retardation. In sickle-cell disease and thalassaemia patients, early detection enables better development and growth [*21–24*].

New forms of secondary prevention, such as genetic or surgical manipulation of an affected fetus to eradicate the biochemical or anatomical abnormality are being tried, some with a high degree of success. This is true for congenital heart disease, cleft lip and cleft palate, congenital dislocation of the hip and others.

Tertiary prevention

Tertiary prevention aims at limiting or reducing the effects of a disorder or disability that is already present. It involves long-term care and management of a chronic condition, e.g. rehabilitation or correction of the disability by surgical measures or by

Table 2 **Usefulness of early detection and intervention programmes**

Disorder	Intervention	Benefit
Hypothyroidism	Hormone replacement therapy	Prevention of mental retardation
Phenylketonuria	Low phenylalanine diet	Prevention of mental retardation
Congential heart disease	Surgical correction	Normal growth and development
Familial growth hormone deficiency	Hormone replacement therapy	Normal growth and development
Familial hypercholesterolaemia	Lowering plasma low-density lipoprotein level by drugs and dietary restriction	Prevention of heart disease
Haemophilia A	Factor VIII replacement	Avoidance of haemorrhage
Infectious diseases:		
poliomyelitis	Vaccination	Avoidance of skeletal disability
meningitis	Vaccination	Avoidance of mental disability
Sickle-cell disease	Proper care and management	Normal growth, decrease in crises
Thalassaemia major	Blood transfusion and iron chelation therapy	Normal growth and development
α_1-antitrypsin deficiency	Avoidance of smoking, α_1-antitrypsin replacement therapy	Delay in lung damage
Congenital dislocation of the hip	Surgical or physical correction	Normal movement
Cleft lip and cleft palate	Surgical correction	Normal facial appearance

adopting strategies by which the disabled person can lead a normal or near normal life. The main aims of rehabilitation of the disabled are:

- to increase awareness of disabilities and the needs of disabled people;
- to encourage their full integration in society; and
- to improve prevention and stimulate a more sensitive and understanding attitude.

These measures also include special education programmes. Only 50 years ago, the majority of disabled people were left illiterate. However, during the past three or four decades, considerable efforts have been made to develop special education programmes to educate blind–deaf–mute, deaf–mute, blind and mentally retarded patients. Special schools with specially trained teachers have provided excellent education programmes which have helped disabled people achieve goals that, in many ways, are similar to those of normal individuals [*20,25*].

Early recognition of disability

To apply any of the previously mentioned preventive measures successfully, the first step is an accurate and early recognition of the disability. Some impairment features are physical and obvious during clinical examination. These include skeletal abnormalities, blindness, hearing and speech disorders, some mental disorders and the chromosomal anomalies such as trisomy 18 or 21, Klinefelter syndrome and Turner syndrome. However, several other disorders do not become evident until later in life, although diagnosis may be made prior to the appearance of the disability or its complications. This has been possible using biochemical tests and, more recently, by applying recombinant DNA technology to the identification of the molecular basis of genetic disability. Examples of tests useful in early diagnosis are shown in Table 1.

The family in general and the mother in particular play a significant role in the early detection of disability. Abnormalities in development, both physical and others, and in learning ability may become obvious to the diligent eyes of the mother much earlier than a clinical diagnosis can be made. Early detection and early intervention can avoid the precipitation of several of the disabilities and can reduce the impact the disability may have on the family.

Once a diagnosis of genetic disorder is made in the carrier parents, proper counselling and premarital testing can prevent the pregnancy of a child with an abnormality. If conception has taken place, then prenatal diagnosis can be used; if the fetus is found to be abnormal, appropriate measures can be adopted and the parents can prepare themselves to look after a disabled child. If the child is diagnosed as having a disease that may lead to a disability, then proper intervention programmes can be started at an early stage and can help ameliorate the effect of the disabling condition. Some examples of the usefulness of early detection and intervention are presented in Table 2.

Medical and/or surgical approaches to preventive intervention provide whatever is necessary to overcome or correct disabilities and strengthen the family unit in order to enhance the abilities of disabled children and their families to cope.

Services and care for disabled people in Saudi Arabia

There are a number of centres of excellence in Saudi Arabia which provide various ser-

vices and care for disabled people. These include specialized disability centres, various medical services at the King Faisal Hospital and Research Centre, university hospitals, military hospitals and Ministry of Health and private hospitals. In addition, special education and psychological and financial support are provided through the Ministry of Labour and Social Welfare, Ministry of Education, General Presidency for Girls' Education, Ministry of Health and the private sector.

The Prince Salman Centre for Disability Research supports research on the prevention, treatment and alleviation of disability in order to improve the quality of life of disabled people and enable them to reach their full potential.

Conclusion

The growing awareness of the problem of disability and the recognition of the need to screen for disability, to classify and assess services available and to optimize these services to meet the needs of disabled people have necessitated a more scientific approach to the problem. The formation of the Saudi Benevolent Association for the Disabled and the subsequent establishment of the Prince Salman Centre for Disability Research and the Prince Salman Bin Abdulaziz Foundation for Humanitarian Services have greatly increased the awareness of such problems in Saudi Arabia. The Ministries of Social Welfare, Health and Education and the Presidency for Girls' Education continue to provide their services to the community, and remarkable improvements have been made.

The national programme for the study of disability nationwide from 1996 to1999 aims at providing a database on the magnitude of the problem and determining its occurrence and distribution and the services available. Assessment of these parameters and recommendations on the appropriateness of approaches for prevention, care and rehabilitation are eagerly awaited.

References

1. McKusick VA. *Mendelian inheritance in man: catalogs of autosomal dominant, autosomal recessive and X-linked phenotypes*, 9th ed. Baltimore, Johns Hopkins University Press, 1990.
2. Contribution of birth defects in infant mortality—United States, 1986. *Morbity and mortality weekly report*, 1989, 38:633–5.
3. La Plante MP. The demographs of disability. In: West J, ed. *The Americans with Disabilities Act: from policy to practice.* New York, Milbank Memorial Fund, 1991: 55–77.
4. Costa T, Scriver CR, Childs B. The effect of Mendelian disease on human health: a measurement. *American journal of genetics*, 1985:231–42.
5. Chen J, Simeonsson RJ. Prevention of childhood disability in the People's Republic of China. *Child care and development*, 1993, 19:71–88.
6. Gortmaker SL, Sappenfield W. Chronic childhood disorders: prevalence and impact. *Pediatric clinics of north America*, 1984, 31:3–18.
7. Hobbs N, Perrin JM, eds. *Issues in the care of children with chronic illness.* San Francisco, Jossey-Bass, 1985:827–63.

8. Cadman D et al. Chronic illness, disability and mental and social well-being: findings of the Ontario Child Health Study. *Pediatrics*, 1987, 79:805–13.

9. Cerreto MC, Travis LB. Implications of psychological and family factors in the treatment of diabetes. *Pediatric clinics of north America*, 1984, 31:689–710.

10. Drotar D, Crawford P, Ganofsky MA. Prevention with chronically ill children. In: Roberts MC, Peterson L, eds. *Prevention of problems in childhood.* New York, John Wiley and Sons, 1984.

11. Breslau N, Salkever D, Staruch KS. Women's labour force activity and responsibilities for disabled dependants: a study of families with disabled children. *Journal of health and social behaviour*, 1982, 23:169–83.

12. Starfield B. Family income, ill health and medical care of US children. *Journal of public health policy*, 1982, 3:244–59.

13. Harper PS. *Practical genetic counselling*, 3rd ed. London, Wright, 1988.

14. Kelly TE. *Clinical genetics and genetic counselling*, 2nd ed. Chicago, Year Book Medical Publishers, 1986.

15. Burnard P. *Counselling skills for health professionals.* London, Chapman and Hall Ltd., 1989.

16. El-Hazmi MAF. Genetic counselling theory and practice. In: *Proceedings of the symposium on medical genetics in the setting of the Middle Eastern population.* Riyadh, KACST Press, 1995:243–6.

17. Emery AE, Pullen I, eds. *Psychological aspects of genetic counselling.* London, Academic Press Inc. Ltd., 1984.

18. Murphy EA, Chase GA. *Principles of genetic counselling.* Chicago, Year Book Medical Publishers, 1975.

19. Angastiniotis M. Social or community response to national control programme for thalassaemia. In: El-Hazmi MAF, ed. *Proceedings of the symposium on blood genetic disorders.* Riyadh, KACST Press, 1994:154–9.

20. Meisels SJ, Shoukoff JP, eds. *Handbook of early childhood intervention.* New York, Cambridge University Press, 1990.

21. Irons M. Screening for metabolic disorders. How are we doing? *Update on neonatology*, 1993, 40:1073–85.

22. American Academy of Pediatrics: newborn screening fact sheet. *Pediatrics*, 1989, 83:449.

23. Abu-Osba YK et al. Comprehensive newborn screening programmes: ARAMCO experience, the national need and recommendations. *Annals of Saudi medicine*, 1992, 12:235–40.

24. Neonatal screening nationwide [Editorial]. *Annals of Saudi medicine*, 1992, 12:233–4.

25. Guralnick MJ, Bennett FC. *The effectiveness of early intervention.* London, Academic Press Inc. Ltd., 1987.