Background information on Health Services for congenital disorders

This document gives a brief overview of Health Services concerned with the care and prevention of congenital disorders. The scope, organisation, staffing and operation of these services vary widely in different countries, depending on many factors including the organisation of Health Services overall, financial resources, demographic and geographic factors (such as the degree of urbanisation), the relationship between the public and private healthcare sectors, and differences in the responsibilities of different types of health professionals. However, it is possible to outline generic features common to effective health care services for these conditions. More detailed information is provided in the documents on preconception, prenatal and newborn services, and in the documents on specific conditions.

The health care pathway for congenital disorders

Health Services for congenital disorders should take a holistic approach to care and prevention that includes:

- Population, public health and environmental health services
- Family planning, women's and reproductive health care services
- Prenatal services
- Maternity services
- Newborn services, including screening for, and diagnosis of, congenital disorders
- Paediatric services, including diagnosis, treatment, care and management
- Life-long medical, social and family support services for those born with congenital conditions.

Services should be well integrated with each other and with other relevant clinical and social services to provide a coherent pathway of care. The establishment and maintenance of effective services requires political commitment, public health and clinical leadership, adequate resources, coordination and teamwork. Professional education and training of the health and social care workforce in both the primary care/community and hospital settings is essential for high quality therapeutic and preventive services. Registers and surveillance systems, to provide epidemiological data and monitor the effectiveness of services and interventions, are important to build a sound evidence base for policy development, planning and action.

Whole-population programmes and interventions

These include:

- Public health programmes to reduce smoking and alcohol consumption
- Fortification of staple foods (for example, with folate or iodine) to reduce the risk of conditions such as neural tube defects and congenital hypothyroidism
- Control of infectious diseases such as syphilis and rubella by education, screening, treatment and (where possible) immunisation programmes
- Environmental health services to combat industrial and agricultural pollution
- Occupational health services to minimise exposure to workplace teratogens.

Effective services require surveillance and monitoring systems (including appropriate laboratory testing services) to assess the scale of environmental hazards such as contamination by specific pollutants (for example, agricultural pesticides) and to measure the burden and spread of infectious diseases. Databases and information systems, supported by appropriate IT infrastructure, should be established to record and collate information. Professionals trained in areas such as public health, toxicology, infectious disease control and occupational health are needed to interpret and analyse this information, and to design suitable programmes for intervention, depending on economic and human resources.

Large-scale programmes such as food fortification, immunisation or control of workplace pollution require leadership, establishment of agreed protocols and procedures, coordination (often at a national level), and in some cases legal or regulatory measures to maximise compliance. All programmes should include resources and procedures for monitoring, quality control and audit.

Interventions before pregnancy

These interventions may be targeted by specific programmes of preconception care for women and couples intending to conceive, and/or more broadly by family planning, women's and reproductive health care services offered to all women of reproductive age. Interventions include:

- Advice and education about the risks of congenital disorders associated with advanced maternal age
- Advice, education and (where possible) behavioural interventions to discourage smoking, excess alcohol consumption and recreational drug use
- Nutritional advice, particularly about folic acid supplementation to reduce the risk of neural tube defects and other conditions
- Advice on genetic risks, including consanguinity, recurrence risk (for example, where there is a family history of a genetic condition, or a previous affected child) and carrier status (for those with a close relative affected by a recessive genetic disease)
- Carrier screening programmes in populations or subpopulations known to have an elevated risk of a specific recessive genetic condition such as sickle cell disease or thalassaemia. These programmes can inform the choice of marriage partner, and/or alert carrier couples to their risk so that they can subsequently, if they wish, opt for prenatal diagnosis
- Sexual health services to control sexually transmitted diseases such as syphilis and HIV-AIDS

- Provision of contraceptives and contraceptive advice to enable women to control family size and spacing, and help minimise the number of births to women of advanced maternal age
- Rubella immunisation.

Family planning, reproductive and women's health clinics may be established as permanent facilities in urban areas, or as outreach clinics in more remote rural areas, particularly in low and middle income countries (LMIC). Basic advice, information and genetic counselling can often be provided by primary healthcare practitioners with appropriate training in the fundamentals of medical genetics: for example in taking and interpreting a family tree and explaining the principles of Mendelian inheritance, including the significance of carrier status for recessive diseases. If resources permit, the service should include provision for access to specialist clinical professionals, for example clinical geneticists, for advice and referral of more complex cases. Programmes such as carrier screening and sexual health services can also be provided in a primary care/community setting, with centralised laboratory testing services and systems to record, track and store samples and results. These programmes are usually established and run at a national or regional level and should have clear protocols and procedures.

Interventions during pregnancy

Where possible, prenatal services should include a specified programme of clinic and/or community outreach visits throughout the pregnancy, in which all aspects of maternal and fetal health are monitored and appropriate advice and interventions are offered. Key components of prenatal care and screening to prevent congenital disorders include:

- Optimising maternal diet; in addition to overall nutrition, supplementation with folic acid, iron and iodine may be considered
- Minimising fetal exposure to teratogens such as tobacco, alcohol and recreational drugs through advice and (if feasible) targeted interventions to help women reduce risky behaviours
- Screening for syphilis, followed by antibiotic treatment of infected women and their partners
- Screening for and management of gestational diabetes
- Screening for Rhesus blood group incompatibility, followed by immunoprophylaxis for Rhesus-negative mothers
- Ultrasound and maternal serum screening programmes to detect Down's syndrome, open neural tube defects and other serious structural anomalies
- Carrier screening in populations with an elevated risk of recessive genetic conditions such as haemoglobin disorders
- Prenatal diagnostic services for genetic conditions. Diagnosis involves amniocentesis or chorionic villus sampling to sample fetal cells, followed by biochemical, cytogenetic or molecular genetic tests to identify specific single-gene or chromosomal disorders. Diagnostic services are offered to women who are known to be at elevated risk of having a child affected by a genetic condition, either because of a family history of a specific condition, a previous affected child, advanced maternal age, or a screen-positive result from a prenatal serum or carrier screening test
- Services for termination of pregnancy (where this is legal and acceptable to the parents) for severe congenital disorders.

Many aspects of routine prenatal care in low-risk women (for example, general advice on diet, smoking or alcohol consumption) may be provided by primary care practitioners such as nurses or other community health workers, supported by primary care doctors and specialist midwives for routine tests and physical examinations.

Prenatal screening and diagnostic services require additional resources such as equipment (for example ultrasound equipment); testing kits (for example, for diabetes or infectious diseases); laboratory services (for example, biochemical laboratories for serum screening, and cytogenetics and molecular genetics laboratories for prenatal genetic diagnosis), expertise (for example, radiographers for ultrasonography, obstetricians for amniocentesis or CVS, practitioners with training in genetic counselling or medical genetics to interpret diagnostic test results and provide genetic information and advice) and infrastructure (for example, systems for sample handling, recording and storing results). These services may be provided in or through dedicated prenatal clinics, often located within hospitals in urban settings.

Where termination of pregnancy is legal in cases of severe fetal abnormality, terminations must be carried out in suitable premises by medically qualified professionals. Resources should be allocated to provide emotional support and counselling to couples throughout the process of diagnosis and decision-making and after the termination has been carried out.

Registers (with appropriate precautions to preserve confidentiality) should be maintained to record numbers of prenatally diagnosed congenital disorders and numbers of terminations for different types of disorder.

Newborn services

The main aim of newborn services in relation to congenital disorders is to diagnose these conditions as promptly as possible, so that appropriate care and treatment can be initiated. The crucial components of neonatal services are diagnostic and screening services within the first few days after birth, and follow-up services during the first few months of life. In many LMIC, where large distances and poverty make follow-up difficult, it is important to ensure that services in the perinatal period are as effective as possible, both to optimise care and management for affected infants, and to ensure that data on affected births (including stillbirths) are recorded.

Newborn services include:

- Provision for all newborns to be examined clinically by a trained healthcare practitioner, both as soon as possible after birth, and within the first few months of life, to detect conditions such as congenital heart defects, dysmorphologies and congenital dislocation of the hip
- Newborn bloodspot or cord blood screening to detect a range of haematological, biochemical and hormonal disorders (many of which are genetic). These include congenital hypothyroidism, phenylketonuria, G6PD deficiency, sickle cell disease and galactosaemia
- Screening for common congenital conditions that are not detected by physical examination, such as hearing defects and congenital cataracts
- Referral of infants with diagnosed or suspected congenital disorders to appropriate clinical specialists.

A fundamental resource for perinatal and neonatal care is the presence of a skilled birth attendant who can help the mother through labour, increase the likelihood of a safe delivery, and perform an initial physical examination of the newborn to ensure prompt recognition of any overt congenital anomalies. Basic maternity and neonatal services may be provided in a community or district hospital setting. The skilled birth attendant may be a doctor, qualified midwife or (in very low resource countries) a community healthcare worker with a more basic level of training.

A more detailed physical examination should be carried out soon after birth, ideally by a primary care doctor or paediatrician trained to recognise congenital disorders and, where possible, initiate appropriate clinical management. When a genetic condition is diagnosed in an infant, services should include provision for follow-up of the family, with the aim of advising couples about the risk to future pregnancies, and identifying and advising other family members who may be at risk. This approach is sometimes called cascade testing or screening.

If more specialist expertise is required, services should include provision for referral to clinicians such as medical geneticists, paediatricians, dieticians, cardiologists or neurologists, for expert examination, additional diagnostic tests (if required), definitive diagnosis, and decisions on clinical management. These services are likely to be established in larger, teaching hospitals in urban areas.

Neonatal screening programmes, like other screening programmes, should be resourced, led and coordinated at a national or regional level, with agreed protocols and standards for testing and reporting results, quality control and audit. Biochemical genetic laboratory services, which will usually be centralised, are required for testing bloodspot samples and storing samples in case of future need. Specialist equipment and trained staff are needed for otoacoustic emission testing and auditory brain stem response for detecting hearing defects. The latter technologies, although currently in use mainly in high income countries, are relatively inexpensive and their application in LMIC is expected to increase.

Wherever possible, resources should be allocated to establishing and maintaining surveillance systems and registers to collect information about the birth prevalence of congenital disorders.

Diagnosis, care and treatment in childhood

Some congenital disorders are not clinically obvious at birth but present during infancy or childhood. Examples include haemophilia, some heart defects and cystic fibrosis. Services should include routine childhood health checks by primary care health workers who are trained to recognise possible birth defects and able to refer cases to specialists for additional investigation if necessary.

For many congenital disorders, effective care can prevent substantial mortality and morbidity. Even where no curative treatment is available, appropriate clinical management, together with social and family support, can make an immense difference to the life of the affected person and the wellbeing of their family, for example by enabling a person who would otherwise be severely disabled to live an independent life.

Treatment and care options may include:

- Paediatric surgery for conditions such as some heart defects, neural tube defects and orofacial clefts
- Services providing non-surgical treatments such as dietary or enzyme replacement therapy for metabolic disorders, speech therapy for those with orofacial clefts, intensive physiotherapy for cystic fibrosis, and blood transfusion (together with iron chelation therapy if necessary) for haemoglobin disorders
- Infection control and pain management.

Surgical and non-surgical treatments for congenital disorders vary widely in their resource requirements. Thyroid hormone treatment for congenital hypothyroidism and routine surgery for some orofacial clefts may be relatively straightforward. However, more complex surgical interventions, such as open-heart surgery for some cardiac defects, require advanced surgical facilities and highly trained clinical professionals.

Life-long support and counselling

Individuals and families affected by congenital disorders often require life-long support. Relevant services may include:

- Rehabilitation and physiotherapy services
- Dietary advice
- Advice on avoiding infections
- Social and psychological support to combat stigmatisation and discrimination
- Special educational provision, especially for those with learning disability
- Community services such as supervised/sheltered housing to encourage inclusion in society
- Advice and counselling to parents and other family members on genetic risks
- Provision for transfer from paediatric to adolescent and adult services.

Resources for these services will generally be provided as part of services for the wider population, rather than designated as services for congenital disorders. The professional roles and competencies required, in addition to medical specialists, include physiotherapy, speech and occupational therapy, dietetics, social work, specialist teaching and counselling.

Cost-effectiveness of interventions

The costs and cost-effectiveness of different types of therapeutic and preventive intervention vary widely, both for different conditions and in different populations. Many whole-population and targeted preconception preventive interventions are clearly cost-effective. For example, in the US, folic acid fortification has been estimated to save US$145 million per year in the costs of caring for children born with spina bifida. The cost of salt iodisation is also negligible compared to the cost of caring for individuals with untreated congenital hypothyroidism. In Iran, the cost of premarital carrier screening and prenatal prevention services is significantly less than the US$200 million per year spent on treating thalassaemia in 2000, before the programme was introduced.

Costs of prenatal screening/testing and diagnosis for congenital disorders, with the option of termination of pregnancy if the fetus is affected, have generally been found to be lower than the lifetime costs of care for affected individuals.

The costs of care and treatment for congenital disorders are highly variable. For some seriously disabling but non-life-threatening conditions such as club foot, or cleft lip/palate, the costs of corrective surgery should be set against the huge cost of life-long disability and disadvantage, both to affected individuals and their families. Whereas untreated individuals may be unable to care for themselves, treatment may allow them to live independent lives and become net financial contributors to society.

Treatments for some congenital disorders are extremely expensive; examples include open-heart surgery for complex cardiac defects, and enzyme replacement therapy for congenital metabolic disorders. These interventions are likely to be beyond the reach of most LMIC and are even challenging for many high and middle income countries. Although provision of the best possible care must always be an ethical imperative, the costs of treatment, which often place enormous financial and other burdens on families, have in many cases led to pressure for the provision of preventive services.

For cost-effectiveness cut-off points for different regions of the world, go to the following website http://www.who.int/choice/costs/CER_levels/en/index.html, and for costs for specific items by region and county, go to http://www.who.int/choice/costs/en/.

What are the main ethical legal and social issues (ELSI) to consider?

All health services for congenital disorders should be based on sound ethical principles. Health professionals in many LMIC may struggle to maintain the highest ethical standards, especially when delivering services to large populations with very limited resources. Financial and social deprivation, or social and cultural practices, may constrain people's behaviour and ability to make autonomous choices. It is important to guard against unrealistic or culturally naive expectations with regard to ethical standards. However, it is equally important to resist the idea that they are a luxury reserved for the well-off; such attitudes may serve to perpetuate, justify and even exacerbate inequality.

One of the most pressing ethical issues relevant to policy development in many LMIC is the absence of safe and affordable medical care for many of the most vulnerable children, including those born with a congenital disorder. Other important issues include the ethical basis for public health interventions, combating discrimination and stigmatisation, informed choice and autonomy, and the legal and social acceptability of prenatal diagnosis and termination of pregnancy. For more detailed discussion of these issues, refer to the supporting chapter on ethical, legal and social issues.

In most countries, those from lower socioeconomic groups suffer a disproportionately high burden of ill health and have poorer access to health services. These problems are often particularly acute in LMIC and may be exacerbated by a split between an impoverished state healthcare sector and a private sector accessible only to the wealthy few. As far as possible, health services for congenital disorders should aim to be equally accessible and equal in quality for all sectors of the population.

The ethical basis for state intervention

When public health interventions are targeted at populations rather than individuals, the intrusiveness of the intervention, and any risks associated with it, should be balanced against the likely benefits. Population-level interventions to prevent congenital disorders, such as folate fortification of staple foods or iodisation of salt, are generally considered to be ethically justified because the interventions are safe, and the benefits in terms of preventing death and disability outweigh the disadvantages of curtailing choice for some individuals.

Discrimination and stigmatisation

Many disabled people, particularly those with physical malformations or learning disability, suffer social as well as medical deprivation. Services designed to care for people with congenital disorders should include social support and societal education to combat stigmatisation; legal measures to discourage discrimination in education and employment may also be considered.

Informed choice

Women and couples who are offered preconception or prenatal interventions to avoid the birth of a child with a congenital disorder should have the right to choose whether to accept these interventions, based on provision of sound information on the nature and likely severity of the condition, the intervention procedure and its risks and benefits. In some cases, it may be difficult to achieve an ethical balance between the autonomous rights of parents (for example, to refuse screening or to choose risky behaviours), and the right of children to be born healthy.

Prenatal diagnosis and termination of pregnancy

The legal and social acceptability of termination of pregnancy varies widely in different countries, due in large part to religious differences. In countries where termination of pregnancy in cases of severe fetal abnormality is legal, this intervention should be offered without coercion and with appropriate support and counselling. Where termination of pregnancy is prohibited, there may be widespread use of unsafe illegal services, which carry serious risks to women's health.

KEY REFERENCES

Alwan A, Modell B. *Community control of genetic and congenital disorders.* EMRO Technical Publications Series 24; 1997.

Christianson A, Howson CP, Modell B *March of Dimes global report on birth defects. The hidden toll of dying and disabled children.* March of Dimes Birth Defects Foundation. White Plains, New York; 2006.

Christianson A, Modell B. *Medical genetics in developing countries*. Annu Rev Genomics Hum Genet 2004; 5:219-65.

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