phg hna toolkit | Congenital Disorders

Background information on Congenital Heart Disease and the impact of interventions

This document gives a brief overview about the condition, its epidemiology and specific interventions that may reduce its burden.

What are Congenital Heart Diseases?

Congenital heart diseases (CHD) are developmental abnormalities of the heart's structure and great vessels that are present at birth. Most involve defects in the heart, valve abnormalities or abnormally draining veins and arteries to and from the heart.

What are the main risk factors?

Genetic factors for CHD include chromosomal abnormalities such as Down's syndrome and Turner's syndrome, and single gene defects such as Alagille syndrome and Noonan syndrome.

Maternal and environmental factors increasing the risk for CHD include maternal diabetes mellitus and phenylketonuria (PKU), maternal obesity, febrile illness, influenza and rubella in pregnancy, medications such as trimethoprim-sulphonamide, retinoic acid used for acne, anti-epileptic medications and organic solvents. There is some evidence that excessive alcohol and smoking are also risk factors for CHD.

In the majority of patients, CHD is thought to be due to a combination of genetic and environmental factors though the aetiology of congenital heart disease is largely unknown.

Global epidemiology

Birth prevalence

CHD is the most common congenital disorder present at birth, reported prevalence ranges from 4/1,000 live births to 12/1,000 live births. Regional and ethnic differences in prevalence have been reported, however, the exact cause of this variation is uncertain. Factors that may influence birth prevalence include maternal age, through its association with chromosomal disorders and maternal nutrition e.g. folate. Prevalence estimates contained within PHGDB come from MGDB and concern isolated congenital heart disease, i.e. defects that involve the

cardiovascular system only and exclude those associated with chromosomal disorders, single gene disorders and those associated with other types of malformations.

Mortality

In the absence of diagnosis or care, infants born with severe CHD have a short life expectancy with many affected children dying in infancy. Additionally, CHD can lead to an increased susceptibility to infection which can increase mortality. Diagnosis and cardiac surgery have led to increased survival of infants with CHD in settings where this is available.

Disability and quality of life

Half of all babies born with significant CHD will require immediate surgery after birth, while most of the remainder may require surgery or medication at some point during their childhood. Disability and quality of life is dependent on the specific defect, some individuals may have minimal disability whist others may develop other health problems through the course of their lives.

Reducing prevalence, morbidity and mortality

Figure 1 illustrates the determinants and interventions for CHD as they relate to key stages in life. The main specific interventions are discussed below.

Interventions before pregnancy

Since most cardiac structures develop in the first 7 weeks after conception when women may be unaware of their pregnancy, the preconception period is a crucial time to identify and minimise behaviours and exposures that may increase risk of CHD.

For women who are intending to become pregnant, the risk of congenital rubella syndrome can be reduced by offering testing for immunity to rubella, and vaccination if susceptible. For women at high risk of infection, vaccination may be offered without susceptibility testing.

Women should be advised about risks of tobacco and alcohol consumption and contact with solvents. Those with chronic health conditions such as diabetes or PKU should be advised to adjust their medications and/or eating habits to keep the conditions under optimum control. Advice should also be given about the risks regarding use of other medications including over the counter drugs, retinoic acid for acne, and oral anticoagulant therapy. Overweight and obese women should be advised to follow appropriate weight loss programmes and hypothyroidism should be treated before conception.

There is strong evidence from observational and randomised controlled trials that multivitamins containing folic acid (400 µg) help to prevent CHD[1,2].

Women with a heart defect or who have had a previous child with a heart defect should be assessed for modifiable risk factors and counselling. If there is a family history of CHD, evaluation for underlying genetic conditions including referral to an experienced medical geneticist may be helpful in defining and managing recurrence risk.

At the population level, good peri-conception health can be fostered by improving access to health care, promoting healthy behaviours, educating health professionals about common preventable risk factors for CHD.

[1] Bailey LB, Berry RJ. Folic acid supplementation and the occurrence of congenital heart defects, orofacial clefts, multiple births, and miscarriage. *Am J Clin Nutr*. 2005 81:1213S-1217S

[2] *Moss and Adam's Heart Disease in Infants, Children, and Adolescents*. Arthur J. Moss, Hugh D. Allen – 2008, 7th edition

Interventions during pregnancy

Precautions such as avoiding tobacco and alcohol consumption, avoiding unnecessary medications, and minimising risk of infection should be maintained during pregnancy in order to reduce the risk of CHD in the fetus.

Many congenital abnormalities can be detected in routine ultrasound examinations during pregnancy. Fetal cardiac ultrasound scans can be performed from 13-14 weeks gestation in specialist centres, though the majority of cases are seen between 18-23 weeks of gestation[3]. For women at high risk (for example because of rubella infection during pregnancy, a family history of CHD, diabetes, PKU or detection of a fetal chromosome abnormality), fetal echocardiography can be used before birth to accurately identify heart defects.

Fetal interventions are possible for some conditions. For example, if the fetus has a heart rhythm abnormality the mother can be given medication to restore normal heart rhythm in the fetus.

If CHD is detected before birth, potential complications during delivery can be anticipated, and delivery in a specialist unit with appropriate medical personnel can be arranged. Early detection may also help prepare the family for the emotional strain, expense, and logistical problems of surgery on the newborn, if this is required. In cases of severe CHD, the option of termination of pregnancy may be considered, taking into account legal and religious issues and acceptability to the parents and society.

Interventions after birth

Diagnosis

Newborn physical examination (screening) can help to identify life-threatening CHD before overt symptoms appear. However, in the newborn, the transition from a fetal to a neonatal circulation can mask the clinical manifestations of CHD. A clinical examination at birth and at 6-8 weeks for all infants, with specific cardiac investigations for high-risk children such as those with Down's syndrome, is recommended[4].

Pulse oximetry and echocardiography, in addition to clinical examination, can improve diagnosis of CHD in the newborn period but their cost-effectiveness has not been adequately evaluated. Screening echocardiography is associated with the highest detection rate but is the most costly strategy and has a 5% false positive rate.

Treatment

The treatment of an affected child depends on the type and severity of his or her heart defect. Other factors include the child's age and general health. The main treatment options are cardiac catheterisation and cardiac surgery. Some children may need several catheter or surgical procedures over a period of years, or they may need to take medicines for long periods in their life. Catheter procedures are less invasive than surgery and since recovery may be faster, they have become the preferred way to repair some simple heart defects, such as patent ductus atriosis, atrial septal defect (ASD) and pulmonary valve stenosis[5].

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[3] British Heart Foundation Factfile 2009: Antenatal screening for congenital heart disease [http://www.bhf.org.uk/publications/view-publication.aspx?ps=1000813] accessed 21 April 2011-04-21

[4] Knowles R, Griebsch I, Dezateux C, Brown J, Bull C, Wren C. Newborn screening for congenital heart defects: a systematic review and cost-effectiveness analysis. *Health Technol Assess*. 2005 Nov;9(44):1-152, iii-iv

[5] NHLI: National Heart, Lung and Blood Institute. Congenital Heart Defects. December 2007[http://www.nhlbi.nih.gov/health/health-topics/topics/chd/treatment.html] (accessed 17 March 2012)

Surgery is the most common way to repair many types of heart defect, including the majority of the most complex ones. “Closed” procedures, where the heart itself is not opened, may be used to repair certain defects such as patent ductus arteriosus or coarctation of the aorta. Open-heart surgery involves placing the patient on a cardiopulmonary bypass machine which does the work of the heart and lungs while the heart is operated on. Open-heart procedures are needed to repair openings in the heart with stitches or with a patch, to repair or replace heart valves, or to widen arteries or openings to heart valves. Catheter and surgical procedures may be combined in a hybrid repair approach for certain complex heart defects. Rarely, children born with complex multiple defects may need heart transplants[8].

Other conditions that are likely to develop in children with CHD and which may need medical treatment include congestive heart failure, arrhythmia and pulmonary hypertension.

Care during childhood

Good hygiene including dental hygiene, frequent hand washing, and avoiding crowded settings and contact with people who are ill, can help prevent infections in a child with CHD. The child should have access to routine care and the standard immunisations that are recommended for all children. Additional immunisations, such as the influenza vaccine, may also be needed. Sometimes children with CHD need a higher calorie diet or have special dietary requirements. Infants and children with CHD tend to gain weight more slowly and may have developmental delay, learning disabilities or special educational needs. However, children with less severe conditions or who have been successfully treated by heart surgery or a catheter intervention may be able to participate fully in normal school programmes and other activities.

Implementation strategies that enable patients to continue regular medical follow-up is critical[6] though this may be challenging in LMIC. Heart check-ups are usually scheduled more frequently during the first few months after the diagnosis or surgery and less often later. For minor conditions check-ups may only be needed every one to five years. Depending on the child’s problem, periodic testing may be needed. These tests may include standard electrocardiogram, 24-hour ambulatory electrocardiogram, chest X-ray, routine (transthoracic) echocardiogram, transesophageal echocardiography, MRI or CT scanning of the heart, exercise stress testing, cardiac catheterisation and angiography.

Cost-effectiveness of interventions

Little information is available on either the costs or cost-effectiveness of interventions for CHD, especially in lower-income countries. Estimates from the United States suggest that the lifetime cost in one year for all babies born with major CHDs (single ventricle, tetralogy of Fallot, transposition of the great arteries and truncus arteriosus) is about US$1.2 billion[7]. This figure includes a wide range of indirect costs such as loss of productivity due to early death, as well as the direct costs of treatment. The costs of care and treatment for less severe CHD are likely to be lower but may still be substantial if lifelong care and surveillance are needed. Costs will vary widely in different countries, depending on the types of tests and treatments available.

[6] Moons P, Hilderson D, Van Deyk K. Implementation of transition programs can prevent another lost generation of patients with congenital heart disease. Eur J Cardiovasc Nurs. 2008 Dec;7(4):259-63

[7] Moss and Adam's Heart Disease in Infants, Children, and Adolescents. Arthur J. Moss, Hugh D. Allen – 2008, 7th edition

Issues of cost-effectiveness are also quite specific to each country. For cost-effectiveness cut-off points for different regions of the world, go to 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?

Equity

Prevention of CHD focuses on good preconception information such as on nutrition and infection control in women of child-bearing age, as well as pregnancy and maternal care. Equity of access to food, sanitation and vaccination programmes is important.

For children born with CHD, effective diagnosis, treatment and care (including long-term follow-up) are specialised and very costly. Here, the issue of equity for families with limited financial resources is even more acute, especially in countries without universal access to healthcare.

Social disadvantage

Diagnosis of CHD in an infant or young child places a heavy emotional burden on families, and affected children may encounter social stigmatisation and educational disadvantage.

Termination of pregnancy

If CHD is diagnosed in a fetus by prenatal testing or screening, the question of termination of pregnancy may arise. In countries where termination in cases of fetal abnormality is legal, medical and ethical judgments may need to be made about whether the condition is serious enough to warrant termination. Parents must be free to exercise autonomous choice to continue an affected pregnancy if they wish to do so. Access to termination of pregnancy is illegal or severely restricted in many LMIC. For women resorting to illegal terminations, the medical, legal and social risks are likely to be high.

Confidentiality

Where a familial risk of CHD is present, health professionals must ensure that genetic information is handled sensitively and that confidentiality is maintained.

KEY REFERENCES

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Figure 1: Needs assessment flowchart for congenital heart defects