Clinical Review

Clinical Review identifies issues in the medical literature of interest to clinicians in Africa. Essential references are given at the end of each section

Cardiology

Congenital Heart Disease: A simplified approach for primary care health workers

Congenital heart defects (CHD) are the commonest birth defects. The estimated prevalence of CHD is 8-10 per 1000 live births, with higher rates seen among still births, spontaneous abortions and premature babies.[1] CHD are the commonest causes of heart disease in children. These cardiac defects generally arise either due to aberrant development of a normal fetal cardiac structure or failure of progression beyond the early embryonic or fetal development stages.[2] Advances in diagnosis and surgical or catheter-based treatment options in the past half a century has resulted in dramatic increase in survival and quality of live for children born with significant heart defects. Without intervention, it is estimated that from one third to 50% of all children born with significant CHD in developing countries will die in the first month of life.[3] This makes early recognition of critical importance.

The etiological factor in most CHD remains largely unknown, with multifactorial factors including genetic predisposition and environmental factors thought to play a role. Several chromosomal abnormalities and genetic aberrations are known to cause CHD (see Table 1). During pregnancy, environmental factors such as maternal infections (Rubella), drug intake (alcohol, lithium, thalidomide and hydantoin) and maternal illnesses (diabetes mellitus, systemic lupus erythromatosus and other autoimmune diseases) are known to cause CHD.[4]

Congenital cardiac defects are classified generally into two broad categories: acyanotic or cyanotic defects. Acyanotic defects include shunt lesions (such as ventricular and atrial septal defects, patent duct arteriosus, etc.), obstructive lesions (e.g. pulmonary and aortic stenosis, coarctation of the aorta) and regurgitant lesions (e.g. mitral valve prolapse). Cyanotic heart diseases are divided into lesions with reduced pulmonary blood flow (e.g. tetralogy of Fallot), defects with increased pulmonary blood flow (e.g. transposition of the great arteries) and total mixing lesions (e.g. Persistent Truncus arteriosus).

Clinical presentation

The clinical presentation of a child with CHD will vary depending on the patient's age, type and severity of the defect and the presence of other associated abnormalities. In the newborn period, clinical cues for severe CHD include respiratory distress, cyanosis and poor peripheral pulses. Cyanosis becomes clinically apparent when arterial oxygen saturations drop below 85%, hence it is important to use pulse oximetry to measure oxygen saturations. In the neonatal period, presenting features of CHD may include cyanosis, heart failure, failure to thrive and abnormal findings on the clinical exam (Table 2). Among infants and older children, usual presenting features include cyanosis, digital clubbing, heart murmurs, heart failure, poor weight gain, squatting episodes (Tetralogy of Fallot), syncope, arrythmias and complications such as repeated chest infections.[5] Whereas in most patient's CHD occurs as an isolated

Table 1. Examples of common easily recognisable chromosomal and genetic abnormalities associated with CHD

Common chromosomal or genetic abnormalities	Main clinical findings	Associated cardiac defects
Down's Syndrome	Mental retardation, Hypotonia, low set ears, slit like eyes, up slanting palpebral fissures, brachycephaly, clinodactyly, macroglossia	AVSD
Noonan's syndrome	Short stature, low set ears, webbed neck, scoliosis, pectus carinatum or excavatum	PS
Turner's Syndrome	Female gender, short stature, webbed neck, broad shield like chest, widely spaced nipples	COA, BAV
DiGeorge Syndrome	Receded chin, low set ears, hypertelorism, narrow philtrum, cleft palate	D-TGA, PTA, TOF
Holt-Oram syndrome	Abnormalities of upper limb bones (arms, wrists and hands)	ASD, Conduction abnormalities

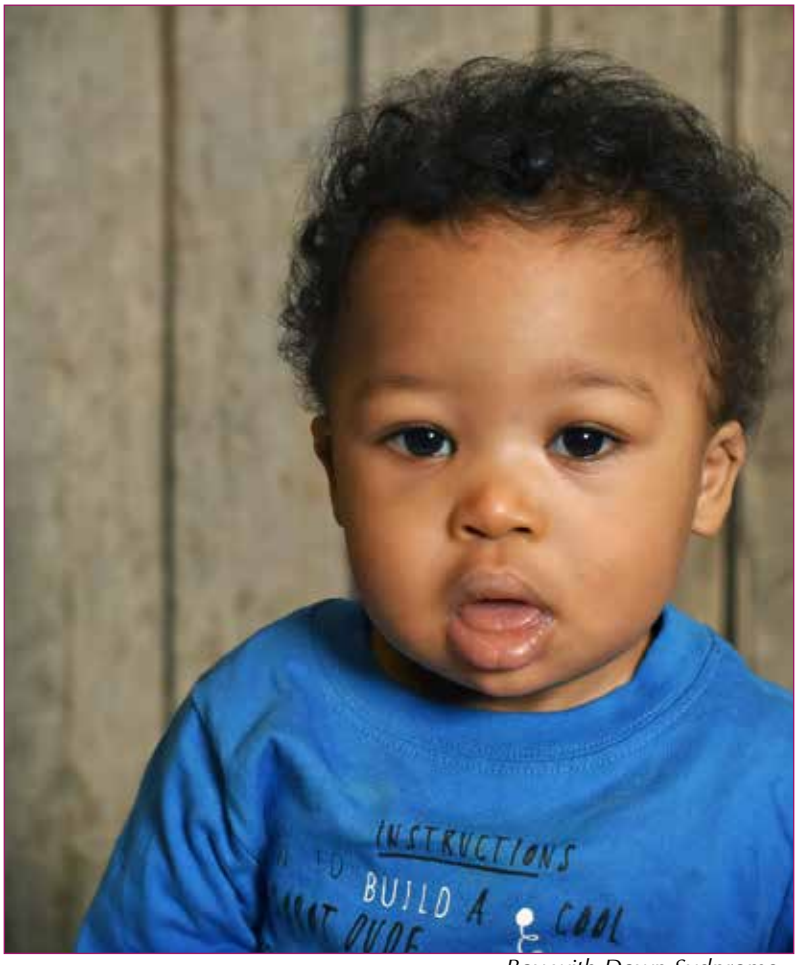
ASD: Atrial septal defect; AVSD; Atrioventricular septal defect; BAV: Bicuspid aortic valve; COA: Coarctation of Aorta; D-TGA: Dextro-transposition of Great arteries; PS: Pulmonary stenosis; PTA: Persistent truncus arteriosus; TOF: Tetralogy of Fallot.

malformation, about one third of cases have associated malformations (see Table 2) and 10-15% have specific genetic syndromes.[2]

Management at primary care facilities

Primary health care workers play an important role in the initial clinical evaluation, identification, stabilisation and referral of patients suspected to have CHD.[6] History should focus on the presence of symptoms such as poor weight gain, recurrent dyspnea and respiratory tract infections that suggest heart failure. Frequent squatting episodes and cyanosis suggests cyanotic CHD such as tetralogy of Fallot. A careful family history spanning at least three generations should be taken, as some defects have higher rates of recurrence and some genetic syndromes associated with CHD have variable expression (2). Clinical examination should focus on identification of a recognisable dysmorphic syndrome and other anomalies often associated with CHD, and the presence of a cardiac murmur (see Table 2). Blood pressure (BP) should be measured in all four limbs, and if the BP in upper limb is greater than that in the lower limbs, this suggests coarctation of the aorta. Pulse oximetry should be performed preferably on the right upper limb and either of the lower limbs, and levels below 92% in room air warrant further cardiac evaluation. Whenever available a chest x-ray should be performed if a child is suspected to have CHD, to evaluate for cardiomegaly. All children with clinical features suggestive of CHD should be referred for echocardiography and cardiologists review.

In addition, primary care health workers can provide surveillance for and initiation of treatment of complications associated with CHD such as Tet spells in Tetralogy of Fallot, treatment of pneumonia and heart failure related to defects with increased pulmonary blood flow (see Table 3).



Boy with Down Sydnrome

Prevention of CHD

Several interventions are possible to reduce morbidity and mortality associated with CHD. Primary preven-

Table 2. Physical findings suggestive of CHD

Clinical exam focus area	Clues suggestive of CHD
General exam	Presence of a recognizable genetic syndrome associated with CHD, cyanosis, digital clubbing, other associated congenital anomalies
Chest exam	Tachypnoea and chest indrawing at rest, pectus carinatum or excavatum
Cardiovascular exam	Bounding pulses, Resting tachycardia, elevated Blood pressure (upper arm>Lower limb), radiofemoral pulse delay, Precordial bulge and hyperactivity, cardiac murmur
Musculo-skeletal system	Limb defects (such as absent radii)
Gastro-intestinal system	Presence of malformations such as Tracheo-esophageal fistulae, Cleft lip/Palate, and anorectal malformations which may be associated with CHD

Table 3. Management of common complications associated with CHD

Complication	How to recognise it	Pre-referral management at primary care facilities
Newborn with critical cyanotic CHD	Severely ill newborn with respiratory distress, visible cyanosis, weak or absent pulses, hypotension	a. Supplemental oxygen b. Cautious Intravenous fluids to maintain appropriate fluid balance c. Counselling of family before referral for cardiology review
Failure to Thrive	Child is small for age, with extreme wasting and stunting	a. Working with family and nutritionists for increased calorific intake
Heart failure	Poor weight gain, recurrent chest infections, diaphoresis, tachypnoea and chest indrawing at rest	a. Appropriate doses of diuretics (furosemide)
Pneumonia	Cough, fever, tachypnoea, crepitations in the lungs	a. Appropriate antibiotics as per local protocol
Tet Spells	Cyanotic child with hyperventilation, worsening cyanosis, irritability, convulsions or coma. Usually triggered by febrile illness or diarrhea with dehydration	a. Placement of the child in knee chest position or its equivalent b. Supplemental oxygen c. Appropriate intravenous fluids d. Oral propranolol (0.5-1mg/kg given 2-3 times daily) e. Check hemoglobin and transfuse with whole blood if Hb<10g/dl
Infective endocarditis	Persistent fevers lasting more than four days despite adequate treatment	a. Do Complete blood count and Blood culture to exclude bacteremia

tion of CHD involves modification and reduction of maternal risk factors associated with CHD. These include measures such as vaccination against Rubella, folic acid supplementation before conception and adequate glycemic control in patients with diabetes. In addition, measures to prevent preterm delivery, such treatment of pregnancy induced hypertension and skilled antenatal care attendance are recommended as Patent ductus arteriosus is a common complication of prematurity. Appropriate nutrition, exercise, and weight control are health promotive behaviors that should be encouraged in prospective mothers. Primary care health workers play a critical role in secondary prevention of CHD which encompasses interventions related to recognition of asymptomatic disease and referral to specialist evaluation for early initiation of treatment before disease progression and development of complications. This includes early referral for definitive surgery or catheter based cardiac interventions. Tertiary prevention involves limiting disability and aiding recovery from complications, through cardiac rehabilitation (7).

Conclusion

Congenital heart defects are the commonest causes of heart disease in children. Without intervention, most children born with significant CHD will die before their first birthday. Primary care health workers play a crucial role in the early identification, initial treatment and referral for definitive diagnosis and treatment of children with CHD.

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