

# CONGENITAL HEART DISEASE IN THE NEWBORN

## Introduction

Congenital heart diseases (CHD) encompass a spectrum of structural abnormalities of the heart or intrathoracic vessels.

Commonly presents in the newborn with central cyanosis, heart failure, sudden collapse or heart murmur.

## Central Cyanosis

- bluish discoloration of lips and mucous membranes
- caused by excess deoxygenated haemoglobin (> 5 Gm/dL)
- confirmed by pulse oxymetry ( $SpO_2 < 85\%$ ) or ABG

## Heart Failure

clinical presentation may mimic pulmonary diseases or sepsis:

- tachypnoea
- hepatomegaly
- tachycardia
- weak pulses

## Sudden Collapse

can be difficult to be distinguished from sepsis or metabolic disorders:

- hypotension
- metabolic acidosis
- extreme cyanosis
- oliguria

Table 2. Causes of heart failure in the newborn

### structural heart lesions

#### obstructive left heart lesions

hypoplastic left heart syndrome, critical aortic stenosis, severe coarctation of aorta

#### severe valvular regurgitation

truncal arteriosus with truncal valve regurgitation

#### large left to right shunts

patent ductus arteriosus, ventricular septal defects, truncus arteriosus, aortopulmonary collaterals

#### obstructed pulmonary venous drainage

total anomalous pulmonary venous drainage

### myocardial diseases

#### cardiomyopathy

infant of diabetic mother, familial, idiopathic ischaemic

anomalous origin of left coronary artery from pulmonary artery, perinatal asphyxia

#### myocarditis

#### arrhythmias

atrial flutter, SVT, congenital heart block

### extracardiac

#### severe anaemia

neonatal thyrotoxicosis

fulminant sepsis

Table 1. Causes of cyanosis in the newborn

### cyanotic heart diseases

#### obstructed pulmonary flow

pulmonary atresia, critical pulmonary stenosis, tetralogy of Fallot

#### discordant ventriculo-arterial connection

transposition of great arteries

#### common mixing

single ventricle, truncus arteriosus, tricuspid atresia, total anomalous pulmonary venous drainage

### primary pulmonary disorders

#### parenchymal disease

meconium aspiration syndrome, respiratory distress syndrome, congenital pneumonia

#### extraparenchymal disease

pneumothorax, congenital diaphragmatic hernia

### persistent pulmonary hypertension of newborn

#### primary

#### secondary

meconium aspiration, perinatal asphyxia, congenital diaphragmatic hernia

### severe polycythaemia

### methaemoglobinaemia

Table 3. Congenital heart lesions that may present with sudden collapse

### duct-dependent systemic circulation

coarctation of aorta, critical aortic stenosis, hypoplastic left heart syndrome, interrupted aortic arch

### duct-dependent pulmonary circulation

pulmonary atresia with intact ventricular septum, tricuspid atresia with pulmonary atresia, single ventricle with pulmonary atresia, critical pulmonary stenosis

transposition of great arteries without septal defect  
obstructed total anomalous pulmonary drainage

## Challenges and Pitfalls

- cyanosis is easily missed in the presence of anaemia.
- difficulty to differentiate cyanotic heart disease from non-cardiac causes
- indistinguishable clinical presentations between left heart obstructive lesions and severe sepsis or metabolic disorders
- possibility of congenital heart disease not considered in management of sick infant

## Clinical Approach to infants with Congenital Heart Disease

### History

- antenatal scans (cardiac malformation, fetal arrhythmias, hydrops)
- family history of congenital heart disease
- maternal illness: diabetes, rubella, teratogenic medications
- perinatal problems: prematurity, meconium aspiration, perinatal asphyxia

### Physical Examination

- dysmorphism: Trisomy 21, 18 and 13, Turner syndrome, DiGeorge syndrome
- central cyanosis
- differential cyanosis (SpO<sub>2</sub> lower limbs < upper limbs)
- tachypnoea
- weak or unequal pulses
- heart murmur
- hepatomegaly

### Investigations

- chest X-ray
- hyperoxia test: administer 100% oxygen via headbox at 15 L/min for 15 mins.  
ABG taken from right radial artery  
cyanotic heart diseases: pO<sub>2</sub> < 100 mmHg; rise in pO<sub>2</sub> is < 20 mmHg  
(note: in severe lung diseases & PPHN, pO<sub>2</sub> can be < 100 mmHg)
- echocardiography

Table 4. Summary of clinical approach to cyanotic newborns

cause	history, signs	chest X-ray	ABG	hyperoxia test	Echo
<b>cyanotic heart disease</b>	no / mild respiratory distress; heart murmur	abnormal heart size, pulmonary vasculature	low pO <sub>2</sub> normal pCO <sub>2</sub>	no rise in pO <sub>2</sub>	usually diagnostic
<b>primary lung disease</b>	respiratory distress	abnormal lungs	low pO <sub>2</sub> high pCO <sub>2</sub>	pO <sub>2</sub> > 100 mmHg	normal
<b>persistent pulmonary hypertension</b>	suggestive history (MAS, asphyxia, sepsis)	may be abnormal (lungs)	differential cyanosis	inconclusive	right to left shunt across PFO or PDA
<b>methemoglobinemia</b>	normal	normal	normal	pO <sub>2</sub> > 100 mmHg	normal

MAS, meconium aspiration syndrome; PFO, patent foramen ovale; PDA, patent ductus arteriosus

### General principles of management

- initial stabilization – secure airway, adequate ventilation, circulatory support
- correct metabolic acidosis, electrolyte, hypoglycaemia; prevent hypothermia
- empirical treatment with IV antibiotics
- early cardiology consultation
- IV Prostaglandin E infusion if duct-dependent lesions suspected  
starting dose: 10 – 40 ng/kg/min; maintenance: 2 – 10 ng/kg/min  
adverse effects: apnoea, fever, hypotension
- unresponsive to IV prostaglandin E, consider:
  - transposition of great arteries, obstructed total anomalous pulmonary venous drainage
  - blocked IV line
  - non-cardiac diagnosis
- arrangement to transfer to regional cardiac center once stabilized