congenital heart diseases

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congenital heart disease, is a problem in the structure of the heart that is present at birth.

Signs and symptoms depend on the specific type of problem. Symptoms can vary from none to life-threatening. When present they may include rapid breathing, bluish skin, poor weight gain, and feeling tired. It does not cause chest pain.

Complications that can result from heart defects include heart failure.

Cause of a congenital heart disease is often unknown. Certain cases may be due to environmental reason. A number of genetic conditions are associated with heart defects including Down syndrome, Turner syndrome, and Marfan syndrome.
Congenital heart defects are divided into two main groups: **cyanotic heart defects** and **non-cyanotic heart defects**, depending on whether the child has the potential to turn bluish in color. The problems may involve the **interior walls of the heart**, the **heart valves**, or the **large blood vessels** that lead to and from the heart.

**Congenital heart defects are partly preventable through**

1. Rubella vaccination
2. Addition of iodine - salt with iodine
3. Addition of folic acid to certain food products.

**Treatment**

Some defects do not need treatment. Other may be effectively treated with catheter based procedures or heart surgery. Occasionally a number of operations may be needed. Occasionally heart transplantation is required. With appropriate treatment outcomes, even with complex problems, are generally good.
Signs and symptoms of congenital heart diseases

Signs and symptoms are related to type and severity of the heart defect. Symptoms frequently present early in life, but it is possible for some heart diseases to go undetected throughout life. Some children have no signs while others may exhibit shortness of breath, cyanosis, fainting, heart murmur, underdevelopment of limbs and muscles, poor feeding or growth, or respiratory infections.

Heart murmur can sometimes be detected by auscultation.

Digital clubbing with cyanotic nail beds in an adult with tetralogy of Fallot
Causes

1- Genetic: Some genes are associated with specific defects. Genetic defect could be due to focal mutations or deletion or addition of segments of DNA.

   a- Large chromosomal abnormalities such as trisomies 21, 13, and 18
   b- Long arm of chromosome 22, 1
   c. Short arm of chromosome 8 (less recurrent regions of the genome)

2- Environmental

Infections during pregnancy: Rubella

Drugs (alcohol, hydantoin, lithium and thalidomide)

Maternal illness (diabetes mellitus, phenylketonuria, and systemic lupus erythematosus).

Maternal obesity

Folate deficiency and diabetes
Mechanism of congenital heart diseases

During development of heart, a complex sequence of events takes place resulting in a well-formed heart at birth. The orderly timing of cell growth, cell migration, and programmed cell death ("apoptosis") has been studied extensively and the genes that control the process are being elucidated. Disruption of any portion may result in a congenital defect.

Diagnosis

*diagnosed prenatally by foetal echocardiography*. This is a test which can be done during the second trimester of pregnancy, when the woman is about 18–24 weeks pregnant. It can be an *abdominal ultrasound* or *transvaginal ultrasound*. After *birth*: by signs and symptoms and through using diagnostic facilities
Classification of congenital heart defects

1- Hypoplasia

Underdevelopment of the right ventricle or the left ventricle. This causes only one side of the heart to be capable of pumping blood to the body and lungs effectively.

It is called **hypoplastic left heart syndrome** when it affects the left side of the heart and **hypoplastic right heart syndrome** when it affects the right side of the heart. In both conditions, there is presence of a patent ductus arteriosus and, when hypoplasia affects the right side of the heart, a **patent foramen ovale**.
2- Obstruction defects
Obstruction defects occur when heart valves, arteries, or veins are abnormally narrow or blocked.
Common defects include pulmonary stenosis, aortic stenosis, and coarctation of the aorta.
Any narrowing or blockage can cause heart enlargement or hypertension.
3- Septal defects

Ventricular septal defects are the most common type of congenital heart diseases, although approximately 30% of adults have a type of atrial septal defect called patent foramen ovale.
4- Cyanotic defects

They result in cyanosis, a bluish-grey discoloration of the skin due to a lack of oxygen in the body. Such defects include persistent truncus arteriosus, total anomalous pulmonary venous connection, tetralogy of Fallot, transposition of the great vessels, and tricuspid atresia.
Tricuspid atresia

Transposition of the great arteries
Clinical Correlates- Septal Defects

1- Atrial septal defect
   a) *Ostium secundum* = excess resorption of septum primum or inadequate development of septum secundum (foramen ovale defect)
   b) *Ostium primum* = septum primum fails to fuse with endocardial cushion (low defect with semilunar shape, right above the AV valves)

2- Ventricular septal defect
   a) *Failure* of membranous portion to develop from extension of endocardial cushion to fuse with interventricular muscular septum
   b) *Muscular defect* = resorption of septum

Clinical Correlates – Conotruncal Septation

1- *Truncus arteriosus* = defective fusion of bulbotruncal ridges
2- *Transposition of Great Arteries* = failure of conotruncal spiral
3- *Tetralogy of Fallot* = unequal division of conus cordis
4- *Semilunar valve stenosis* = failure of development of conotruncal swellings or unequal partition
5- *Patent ductus arteriosus*: failure of closure of the ductus arteriosus
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