CONGENITAL HEART DEFECT(CHD)

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Summary: In this article, I touched on one of the open topics in medicine, like pancreatic cancer. Many people require lifelong specialized cardiac care, first with a pediatric cardiologist and later with an adult congenital cardiologist. There are more than 1.8 million adults living with congenital heart defects.
Keywords: signs and symptoms, changes at birth, hypoplasia.

Briefly about the disease

A congenital heart defect , also known as a congenital heart anomaly and congenital heart disease, is a defect in the structure of the heart or great vessels that is present at birth. A congenital heart defect is classed as a cardiovascular disease. Signs and symptoms depend on the specific type of defect. Symptoms can vary from none to life-threatening.[7] When present, symptoms may include rapid breathing, bluish skin (cyanosis), poor weight gain, and feeling tired. Congenital Heart Defect does not cause chest pain. Most congenital heart defects are not associated with other diseases. A complication of CHD is heart failure.

Signs and symptoms

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 CHDs to go undetected throughout life. Some children have no signs while others may exhibit shortness of breath, cyanosis, fainting, heart murmur, under-development of limbs and muscles, poor feeding or growth, or respiratory infections. Congenital heart defects cause abnormal heart structure resulting in production of certain sounds called heart murmur. These can sometimes be detected by auscultation; however, not all heart murmurs are caused by congenital heart defects.

Causes

The cause of congenital heart disease may be environmental, or a combination of both.

Molecular pathways

The genes regulating the complex developmental sequence have only been partly elucidated. Some genes are associated with specific defects. A number of genes have been associated with cardiac manifestations. Mutations of a heart muscle protein, α-myosin heavy chain (MYH6) are associated with atrial septal defects. Several proteins that interact with MYH6 are also associated with cardiac defects. The transcription factor GATA4 forms a complex with the , which interacts with MYH6. Another factor, the homeobox (developmental) gene, NKX2-5 also interacts with MYH6. Mutations of all these proteins are associated with both atrial and ventricular septal defects; In addition, NKX2-5 is associated with defects in the electrical conduction of the heart and TBX5 is related to the Holt–Oram syndrome which includes electrical conduction defects and abnormalities of the upper limb. The Wnt signaling co-factors BCL9, BCL9L and PYGO might be part of this molecular pathways, as when their genes are mutated, this causes phenotypes similar to the features present in Holt-Oram syndrome.

Another T-box gene, TBX1, is involved in velo-cardio-facial syndrome DiGeorge syndrome, the most common deletion which has extensive symptoms including defects of the cardiac outflow tract including tetralogy of Fallot.

Environmental

Known environmental factors include certain infections during pregnancy such as rubella, drugs (alcohol, hydantoin, lithium and thalidomide) and maternal illness (diabetes mellitus, phenylketonuria, and systemic lupus erythematosus).[Alcohol exposure in the father also appears to increase the risk of congenital heart defects.

Being overweight or obese increases the risk of congenital heart disease. Additionally, as maternal obesity increases, the risk of heart defects also increases. A distinct physiological mechanism has not been identified to explain the link between maternal obesity and CHD, but both pre-pregnancy folate deficiency and diabetes have been implicated in some studies.

Mechanism

There is a complex sequence of events that result in a well formed heart at birth and disruption of any portion may result in a defect. 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. Around day 15 of development, the cells that will become the heart exist in two horseshoe shaped bands of the middle tissue layer (mesoderm), and some cells migrate from a portion of the outer layer (ectoderm),neural crest, which is the source of a variety of cells found throughout the body. On day 19 of development, a pair of vascular elements, the "endocardial tubes", form. The tubes fuse when cells between then undergo programmed death and cells from the first heart field migrate to the tube, and form a ring of heart cells (myocytes) around it by day 21. On day 22, the heart begins to beat and by day 24, blood is circulating.

At day 22, the circulatory system is bilaterally symmetrical with paired vessels on each side and the heart consisting of a simple tube located in the midline of the body layout. The portions that will become the atria and will be located closest to the head are the most distant from the head. From days 23 through 28, the heart tube folds and twists, with the future ventricles moving left of center (the ultimate location of the heart) and the atria moving towards the head.

On day 28, areas of tissue in the heart tube begin to expand inwards; after about two weeks, these expansions, the membranous "septum primum" and the muscular "endocardial cushions", fuse to form the four chambers of the heart. A failure to fuse properly will result in a defect that may allow blood to leak between chambers. After this happens, cells that have migrated from the neural crest begin to divide the bulbus cordis, the main outflow tract is divided in two by the growth a spiraling septum, becoming the great vessels—the ascending segment of the aorta and the pulmonary trunk. If the separation is incomplete, the result is a "persistent truncus arteriosus". The vessels may be reversed ("transposition of the great vessels"). The two halves of the split tract must migrate into the correct positions over the appropriate ventricles. A failure may result in some blood flowing into the wrong vessel (e.g. The four-chambered heart and the great vessels have features required for fetal growth. The lungs are unexpanded and cannot accommodate the full circulatory volume. Two structures exist to shunt blood flow away from the lungs. Cells in part of the septum primum die creating a hole while muscle cells, the "septum secundum", grow along the right atrial side the septum primum, except for one region, leaving a gap through which blood can pass from the right

artium to the left atrium, the foramen ovale. A small vessel, the ductus arteriosus allows blood from the pulmonary artery to pass to the aorta.

Changes at birth

The ductus arteriosus stays open because of circulating factors including prostaglandins. The foramen ovale stays open because of the flow of blood from the right atrium to the left atrium. As the lungs expand, blood flows easily through the lungs and the membranous portion of the foramen ovale (the septum primum) flops over the muscular portion (the septum secundum). If the closure is incomplete, the result is a patent foramen ovale. The two flaps may fuse, but many adults have a foramen ovale that stays closed only because of the pressure difference between the atria.

Theories

Rokitansky (1875) explained congenital heart defects as breaks in heart development at various ontogenesis. Spitzer (1923) treats them as returns to one of the phylogenesis. Krimski (1963), synthesizing two previous points of view, considered congenital heart diseases as a stop of development at the certain stage of ontogenesis, corresponding to this or that stage of the phylogenesis. Hence these theories can explain feminine and neutral types of defects only.

Hypoplasia

Hypoplasia can affect the heart, typically resulting in the underdevelopment of the right ventricle or the . This causes only one side of the heart to be capable of pumping blood to the body and lungs. Hypoplasia of the heart is rare but is the most serious form of CHD. 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, the presence of a patent ductus arteriosus (and, when hypoplasia affects the right side of the heart, a patent foramen ovale) is vital to the infant's ability to survive until emergency heart surgery can be performed, since without these pathways blood cannot circulate to the body (or lungs, depending on which side of the heart is defective). Hypoplasia of the heart is generally a cyanotic heart defect.

Treatment

CHD may require surgery and medications. Medications include diuretics, which aid the body in eliminating water, salts, and digoxin for strengthening the contraction of the heart. This slows the heartbeat and removes some fluid from tissues. Some defects require surgical procedures to restore circulation back to normal and in some cases, multiple surgeries are needed.

Interventional cardiology now offers minimally invasive alternatives to surgery for some patients. The Melody Transcatheter Pulmonary Valve (TPV), approved in Europe in 2006 and in the U.S. in 2010 under a Humanitarian Device Exemption (HDE), is designed to treat congenital heart disease patients with a dysfunctional conduit in their right ventricular outflow tract (RVOT). The RVOT is the connection between the heart and lungs; once blood reaches the lungs, it is enriched with oxygen before being pumped to the rest of the body. Transcatheter pulmonary valve technology provides a less-invasive means to extend the life of a failed RVOT conduit and is designed to allow physicians to deliver a replacement pulmonary valve via a catheter through the patient's blood vessels.

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