

EMBRYONAL HEART DEFECTS (CONGENITAL HEART DEFECTS)

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Annotation: *This article reviews the main pathologies of congenital heart defects. It discusses the morphological and clinical features, diagnostic approaches, and treatment strategies of heart defects such as Tetralogy of Fallot (TOF), tricuspid atresia, Ebstein's anomaly, Eisenmenger syndrome, transposition of the great arteries, and truncus arteriosus. Additionally, the article examines the embryological mechanisms underlying these defects and their impact on health. It is intended as a theoretical and practical resource for medical professionals and researchers to understand and manage congenital heart defects.*

Keywords: *Congenital heart defects, Tetralogy of Fallot (TOF), Tricuspid atresia, Ebstein anomaly, Eisenmenger syndrome, Transposition of the great arteries, Truncus arteriosus, Embryonic heart development.*

The heart is the first organ to develop among embryonic organs, arising from the mesodermal layer. It appears during the 18th–19th days of embryonic development and begins to loop from the right to the left side by days 21–22. This looping process is regulated by the dynein protein. If this protein is insufficient or absent, Kartagener syndrome may occur, resulting in the heart being positioned on the right side of the thoracic cavity.

Kartagener syndrome is a rare disorder caused by hereditary dysfunction of cytoplasmic cilia and flagella, often associated with Primary Ciliary Dyskinesia (PCD). The main clinical features of the disease include the following:

Firstly, bronchial and pulmonary manifestations: patients commonly present with chronic cough, excessive sputum production, chronic sinusitis, and bronchitis. Over time, bronchial dilatation may develop, leading to bronchiectasis.

Secondly, cytoplasmic and anatomical features: approximately half of affected patients exhibit situs inversus totalis, meaning that the internal organs are mirrored from their usual positions. Consequently, the heart is often located on the right side (dextrocardia).

Thirdly, reproductive system manifestations: in males, infertility arises due to immotile spermatozoa, while in females, dysfunction of the fallopian tubes may impair fertility. Additionally, recurrent ear and nasal infections are frequently observed in these patients.

The clinical features of Kartagener syndrome are largely associated with ciliary dysfunction during embryonic development, which can also lead to abnormal positioning of the heart and other organs. During the fourth week of embryogenesis, the heart chambers begin to form, dividing into atria and ventricles, and rhythmic contractions start. If the spiral looping of the truncal and bulbar ridges is disrupted, Transposition of the Great Arteries (TGA) may occur. In this condition, the outflow tracts of the pulmonary artery and aorta are switched: the pulmonary artery arises from the left ventricle, and the aorta arises from the right ventricle. As a result, oxygen-rich blood is delivered to the right ventricle instead of the left, leading to hypoxia in tissues and organs. In such patients, short embryonic shunts, such as the ductus arteriosus, may remain open or be pharmacologically maintained with prostaglandin E to allow for mixing of oxygenated and deoxygenated blood.

Another congenital heart defect can arise in the presence of **pulmonary vein stenosis**, leading to **Tetralogy of Fallot (TOF)**. TOF results from the involvement of four anatomical abnormalities. The primary cause is initially reduced pulmonary blood flow due to pulmonary stenosis, which may develop as a consequence of limited fetal movement. Over time, compensatory mechanisms increase blood flow, but the stenotic pulmonary veins cannot adequately transport blood, resulting in **right ventricular hypertrophy**. The increased pressure within the right ventricle may damage the membranous portion of the interventricular septum, causing a

ventricular septal defect (VSD). Consequently, the **aorta is displaced anteriorly**, and in some patients, aneurysm formation may also occur.

Total Anomalous Pulmonary Venous Return (TAPVR) is a rare congenital heart defect in which all pulmonary veins fail to connect correctly to the left atrium. Instead, oxygenated blood from the lungs is redirected to the right atrium or other venous pathways, causing mixing of oxygenated and deoxygenated blood.

TAPVR is classified into several types:

1. **Supracardiac TAPVR** – pulmonary veins drain into the superior venous system above the heart; this is the most common type.
2. **Infracardiac TAPVR** – pulmonary veins drain into veins below the diaphragm, often associated with pulmonary vein stenosis.
3. **Cardiac TAPVR** – pulmonary veins drain directly into the right atrium or coronary sinus.
4. **Mixed TAPVR** – a combination of the above pathways.

From a pathophysiological perspective, the misrouting of oxygenated blood leads to mixing of systemic and pulmonary circulation. This results in increased pulmonary pressure and excessive cardiac workload. In cases with venous stenosis, symptoms manifest rapidly after birth.

Clinical manifestations include cyanosis (bluish discoloration of the skin and lips) observed shortly after birth, rapid fatigue, difficulty feeding, recurrent respiratory infections (bronchitis, pulmonary infections), pulmonary and cardiac enlargement, and occasionally heart failure.

Diagnosis primarily relies on echocardiography, which helps identify the abnormal connection of the pulmonary veins. Additionally, chest X-ray may reveal pulmonary enlargement or changes in cardiac silhouette. In complex cases, cardiac catheterization is employed to measure blood flow and intracardiac pressures. Management of TAPVR is exclusively surgical. The goal of the procedure is to redirect all pulmonary veins to the left atrium and restore normal circulation.

Surgery is recommended within the first 1–2 weeks after birth or before the age of one year, as prolonged delay can lead to excessive pulmonary and cardiac workload. The prognosis is generally favorable, with most patients leading normal lives postoperatively; however, long-term follow-up is essential. The presence of venous stenosis or additional congenital heart defects may slightly worsen the prognosis.

Ebstein anomaly is a rare congenital heart defect in which the tricuspid valve is abnormally developed and displaced toward the right atrium. This results in enlargement of the right atrium, reduction of the functional right ventricle, and impaired blood flow. Improper closure of the tricuspid valve leads to tricuspid regurgitation and increased volume load on the right side of the heart.

Clinical features include cyanosis (bluish discoloration of the skin and lips), rapid fatigue, dyspnea, signs of chronic heart failure, and arrhythmias. Diagnosis primarily relies on echocardiography to assess the position of the tricuspid valve and the degree of regurgitation. Electrocardiography (ECG) helps detect arrhythmias, which may occasionally be associated with Wolff-Parkinson-White (WPW) syndrome. Chest X-ray can demonstrate enlargement of the right atrium. Management depends on the severity of the condition. Mild forms may only require monitoring and medical therapy, whereas severe cases necessitate surgical repair or replacement of the tricuspid valve. Prognosis depends on symptom severity and the extent of valvular dysfunction. Patients with mild forms can often lead normal lives, while severe cases require surgical intervention and long-term follow-up.

Eisenmenger syndrome is a chronic cardiovascular condition that develops as a consequence of congenital heart defects, typically involving left-to-right shunts (e.g., ventricular septal defect [VSD], atrial septal defect [ASD], or patent ductus arteriosus [PDA]) that remain untreated over time. These defects increase pulmonary artery pressure, eventually reversing the direction of blood flow. As a result,

oxygenated blood cannot reach systemic circulation, leading to cyanosis and systemic hypoxia.

Etiology: The primary causes of Eisenmenger syndrome are congenital heart defects. The most common are VSD, ASD, and PDA, sometimes in combination with complex defects. These abnormalities lead to increased pulmonary vascular resistance over time, resulting in reversed blood flow and the development of the syndrome. Eisenmenger syndrome includes chronic cyanosis, fatigue, dyspnea, tachycardia, lower extremity edema, and occasionally recurrent pulmonary infections. The syndrome often manifests in adulthood, as it develops gradually due to progressive pulmonary hypertension.

Management: Complete reversal of Eisenmenger syndrome is challenging; therefore, prevention and early correction of congenital heart defects are crucial. Medical therapy aims to alleviate symptoms, reduce cardiac and pulmonary workload, and control blood pressure. In severe cases, heart-lung transplantation or complex surgical interventions may be required.

Atrial Septal Defect (ASD) is a congenital heart defect characterized by a hole in the septum separating the right and left atria. This results in mixing of systemic and pulmonary circulation, as oxygen-rich blood from the left atrium flows into the right atrium. Over time, this may increase pulmonary arterial pressure and impose an excessive workload on the right side of the heart.

Etiology: ASD typically arises from abnormal embryonic development. Incomplete closure of the septum primum and septum secundum is the primary cause. Genetic factors, certain inherited disorders, and maternal exposure to specific drugs or infections during pregnancy may also contribute to ASD formation.

Clinical manifestations: Small ASDs are often asymptomatic and may only be detected in adulthood. Larger defects can present with chronic fatigue, dyspnea, tachycardia, and occasionally cyanosis. Long-standing elevated pulmonary pressure can eventually lead to Eisenmenger syndrome.

Diagnosis: Echocardiography is the primary diagnostic tool, allowing visualization of the septal defect and assessment of blood flow. Electrocardiography (ECG) and chest X-ray may assist in detecting right atrial enlargement or arrhythmias.

Treatment: Small ASDs that are asymptomatic usually require only monitoring. In cases of large ASDs or when symptoms are present, surgical closure or transcatheter device closure of the septal defect is recommended. The primary goal of treatment is to normalize pulmonary arterial pressure and reduce excessive cardiac workload. **Prognosis:** If detected and treated early, patients can lead normal lives. Delayed intervention increases the risk of elevated pulmonary pressure and the development of Eisenmenger syndrome.

Atrial Septal Defect (ASD) is a congenital heart defect characterized by a hole in the septum separating the right and left atria. This results in mixing of systemic and pulmonary circulation, as oxygen-rich blood from the left atrium flows into the right atrium. Over time, this may increase pulmonary arterial pressure and impose an excessive workload on the right side of the heart. **Etiology:** ASD often arises from developmental abnormalities during embryogenesis. Incomplete closure of the septum primum and septum secundum is the primary cause. Genetic factors, certain inherited disorders, and maternal exposure to specific drugs or infections during pregnancy may also contribute to ASD formation.

Clinical manifestations: Small ASDs are frequently asymptomatic and may only be discovered in adulthood. Larger ASDs can present with chronic fatigue, dyspnea, tachycardia, and occasionally cyanosis. Persistent pulmonary hypertension may eventually lead to Eisenmenger syndrome.

Diagnosis: Echocardiography is the primary diagnostic tool, enabling visualization of the septal defect and assessment of blood flow. Electrocardiography (ECG) and chest X-ray may help detect right atrial enlargement or arrhythmias. **Treatment (summary):** Asymptomatic small ASDs are managed with

observation. Large or symptomatic ASDs require surgical or catheter-based closure, with the goal of normalizing pulmonary pressure and reducing cardiac overload. Prognosis: If detected and treated early, patients can lead normal lives. Delayed intervention increases the risk of elevated pulmonary arterial pressure and the development of Eisenmenger syndrome.

Patent Ductus Arteriosus (PDA) is a congenital heart defect in which the ductus arteriosus fails to close after birth, maintaining an abnormal connection between the pulmonary artery and the aorta. Normally, the ductus arteriosus closes within the first few days of life, but in PDA it remains patent, allowing oxygen-rich blood to mix between the aorta and the pulmonary artery.

Etiology: PDA commonly results from developmental abnormalities during embryogenesis. Contributing factors include maternal infections, drug exposure during pregnancy (e.g., ibuprofen or indomethacin), prematurity, genetic predisposition, and intrauterine hypoxia, which can delay closure of the ductus arteriosus. Clinical manifestations: Small PDAs are often asymptomatic. Large PDAs may present with dyspnea, fatigue, frequent pulmonary infections shortly after birth, tachycardia, and occasionally cyanosis. If untreated over a long period, PDA can increase pulmonary arterial pressure and impose excessive workload on the heart.

Diagnosis: Echocardiography is the primary diagnostic tool, allowing visualization of the patent ductus arteriosus and assessment of blood flow. Chest X-ray and ECG may reveal right-sided cardiac enlargement or arrhythmias. Management: Asymptomatic small PDAs may only require monitoring. Large or symptomatic PDAs are treated with medical therapy (e.g., prostaglandin inhibitors), catheter-based closure, or sometimes surgical intervention. The main goal is to normalize pulmonary arterial pressure and reduce cardiac overload.

Prognosis (summary): Early detection and treatment allow patients to lead normal lives. Delayed intervention increases the risk of pulmonary hypertension, chronic heart failure, or the development of Eisenmenger syndrome.

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