Tetralogy of fallot.

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ANNOTATION

Tetralogy of Fallot is the most common cyanotic congenital heart disease and is characterized by a combination of four anatomical abnormalities: ventricular septal defect, pulmonary stenosis, overriding aorta, and right ventricular hypertrophy. These structural defects result in decreased pulmonary blood flow and mixing of oxygenated and deoxygenated blood, leading to chronic hypoxemia and cyanosis. Clinical manifestations usually appear in early infancy and include cyanosis, dyspnea, failure to thrive, and characteristic "tet spells." Advances in diagnostic techniques, particularly echocardiography, allow for early detection of the condition. Surgical correction remains the definitive treatment and has significantly improved survival rates and long-term outcomes. Early diagnosis and timely intervention are crucial for reducing morbidity and mortality associated with Tetralogy of Fallot.

Keywords:

Tetralogy of Fallot, congenital heart disease, cyanosis, ventricular septal defect, pulmonary stenosis, pediatric cardiology

Introduction.

Tetralogy of Fallot (TOF) is a complex congenital cardiac anomaly first described by Étienne-Louis Arthur Fallot in 1888. It accounts for approximately 7–10% of all congenital heart defects. The pathophysiology of TOF is primarily determined by the degree of right ventricular outflow tract obstruction, which influences the severity of cyanosis and clinical symptoms. The severity of cyanosis in TOF depends on the degree of right ventricular outflow tract obstruction. When obstruction is severe, a large right-to-left shunt occurs, causing systemic desaturation and cyanosis. Episodes of acute

cyanosis, known as “tet spells,” often occur during crying, feeding, or exercise due to increased right-to-left shunting. Clinically, TOF presents with cyanosis, dyspnea, fatigue, poor growth, and clubbing of the fingers in chronic cases.

A harsh systolic murmur is commonly detected due to pulmonary stenosis. Early diagnosis is crucial and is primarily achieved through echocardiography, which allows visualization of the four characteristic defects. Electrocardiography may show right ventricular hypertrophy and right axis deviation, while chest X-ray can reveal a classic “boot-shaped” heart. Advanced imaging modalities such as cardiac MRI or CT are helpful for surgical planning and detailed anatomical assessment.

The etiology of TOF is multifactorial. While most cases are sporadic, approximately 10–20% are associated with genetic syndromes such as 22q11.2 deletion (DiGeorge syndrome) and Down syndrome. Environmental factors, including maternal diabetes, phenylketonuria, or exposure to teratogenic drugs during pregnancy, can also contribute to the development of TOF.

Management of TOF involves both medical and surgical approaches. Medical therapy is mainly supportive, aimed at managing tet spells through oxygen supplementation, fluid management, and beta-blockers. Definitive treatment is surgical repair, which typically involves closure of the VSD and relief of right ventricular outflow tract obstruction. Complete repair is usually performed between 3 and 12 months of age. In unstable neonates or those with severe cyanosis, palliative procedures such as the Blalock-Taussig shunt may be performed to increase pulmonary blood flow temporarily.

Surgical advances, including transannular patch repair, valve-sparing techniques, and minimally invasive approaches, have significantly improved survival rates. Lifelong follow-up is essential to monitor for potential complications, including pulmonary regurgitation, arrhythmias, residual VSD, and right ventricular dysfunction. With early

diagnosis and timely surgical intervention, over 85–90% of children with TOF survive into adulthood with a good quality of life.

Tetralogy of Fallot remains a significant cause of morbidity in pediatric populations, but advancements in diagnostic techniques, surgical repair, and long-term care have transformed the prognosis for affected patients. Multidisciplinary management, involving pediatric cardiologists, cardiothoracic surgeons, and specialized nursing care, is critical for optimizing outcomes and preventing long-term complications.

The four classical components of Tetralogy of Fallot include a large ventricular septal defect, obstruction of the right ventricular outflow tract, an overriding aorta that receives blood from both ventricles, and compensatory right ventricular hypertrophy. These abnormalities lead to right-to-left shunting of blood and reduced oxygen saturation in systemic circulation. Tetralogy of Fallot is the most common cyanotic congenital heart disease, representing 7–10% of all congenital heart defects. It is defined by the simultaneous presence of four structural abnormalities: ventricular septal defect, pulmonary stenosis, overriding aorta, and right ventricular hypertrophy. TOF typically presents in infancy with cyanosis and may lead to significant morbidity if left untreated. Advances in surgical repair and perioperative care have significantly improved survival rates, allowing most patients to reach adulthood.

Clinical features vary depending on the severity of pulmonary stenosis and may range from mild cyanosis to severe hypoxic episodes known as “tet spells.” Diagnosis is mainly established through echocardiography, while additional investigations such as electrocardiography, chest radiography, and cardiac MRI provide supportive information.

Definitive management involves surgical repair, typically performed in early childhood, consisting of closure of the ventricular septal defect and relief of right ventricular outflow obstruction. Long-term follow-up is essential, as patients may develop arrhythmias, pulmonary regurgitation, or right ventricular dysfunction later in life.

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