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Pulmonic Stenosis-Ventricular Septal Defect (Teratology of Fallot) Origin and Treatment

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ABSTRACT

Tetralogy of Fallot (TOF) is the most prevalent form of cyanotic congenital heart disease, comprising four distinct anatomical defects with varying degrees of severity. The clinical presentation in neonates largely depends on the extent of right ventricular outflow tract (RVOT) obstruction and the patency of the ductus arteriosus. Common symptoms include irritability, excessive sweating, cyanotic spells, and heart failure. Accurate diagnosis requires a combination of imaging techniques such as prenatal sonography, Doppler echocardiography, cardiac catheterization, multi-detector CT (MDCT), and MRI, all of which provide insights into the complex physiological abnormalities associated with TOF.

Keywords: *Heart diseases, Teratology of Fallot, diagnosis, treatment*

1. INTRODUCTION

Tetralogy of Fallot (TOF) is a congenital anomaly of the heart that interferes with the normal flow of blood due to the presence of four cardinal structural defects. These include a ventricular septal defect (VSD), pulmonary stenosis, right ventricular hypertrophy, and an overriding aorta. This condition develops during fetal life, and its physiological impact results from the mixing of oxygen-poor and oxygen-rich blood, thereby reducing the amount of oxygen available to the body. As a result, TOF is classified as a critical congenital heart defect, often necessitating prompt surgical intervention after birth (1).

The hallmark feature of TOF is cyanosis, a bluish discoloration of the skin caused by insufficient oxygen in the bloodstream. However, this symptom may not be immediately evident in the neonatal period. In many cases, cyanosis becomes more apparent after the closure of the ductus arteriosus, typically within the first days of life. Some infants may present with “tet spells,” which are sudden, severe episodes of cyanosis triggered by activities such as crying or feeding. These spells are characterized by difficulty breathing, irritability, and profound cyanosis, and if not treated urgently, may progress to loss of consciousness, seizures, or even death (2,3).

The clinical severity and timing of symptom onset depend largely on the extent of right ventricular outflow tract (RVOT) obstruction. Despite the variability in presentation, most cases are diagnosed early in life through a combination of clinical assessment and imaging. This early detection has been made possible through improved diagnostic modalities and heightened clinical awareness.

This review provides a comprehensive examination of the underlying causes, clinical features, diagnostic approaches, and current treatment strategies for Tetralogy of Fallot, with the aim of enhancing early diagnosis and improving long-term outcomes for affected individuals.

2. ETIOLOGY AND RISK FACTORS

The exact causes behind the development of Tetralogy of Fallot are not yet fully understood. However, the condition is believed to result from a multifactorial interplay of genetic and environmental influences that affect fetal heart development. Chromosomal abnormalities and mutations in specific genes regulating cardiac formation can predispose a fetus to TOF (1,4).

Certain environmental exposures during pregnancy further increase the likelihood of congenital heart defects. These include maternal infections, particularly viral illnesses such as rubella (German measles), which have been linked to heart malformations. Additional risk factors include maternal alcohol consumption, poor prenatal nutrition, uncontrolled diabetes, and advanced maternal age (typically over 40 years) (5). There is also a higher incidence of TOF among infants born to

mothers who smoke or take certain medications during pregnancy (5).A positive family history of congenital heart disease increases the risk, indicating a possible hereditary component. Furthermore, TOF may occur in association with genetic syndromes such as Down syndrome and DiGeorge syndrome, both of which involve chromosomal deletions or anomalies that affect organogenesis (6).

Recent studies have identified mutations in genes such as NKX2-5, JAG1, and GATA4 as being associated with conotruncal defects, including TOF. These gene mutations disrupt normal cardiac morphogenesis and contribute to the heterogeneity of congenital heart anomalies (7). Genetic testing and counseling may be advised in families with a history of congenital cardiac malformations. One significant complication of TOF is the increased susceptibility to infective endocarditis, a bacterial infection that targets the inner lining of the heart or its valves. For this reason, patients with TOF are often advised to take prophylactic antibiotics before undergoing dental or certain surgical procedures to reduce the risk of infection. If left untreated, TOF can result in serious long-term complications, including disability or premature death in early adulthood (1).

3. SIGNS AND SYMPTOMS OF FALLOT

Tetralogy of Fallot is commonly recognized and diagnosed within the first few weeks after birth, often prompted by the presence of a loud cardiac murmur or signs of cyanosis. In many newborns, a patent ductus arteriosus (PDA) remains open immediately after birth, allowing additional blood flow to the lungs. This temporary compensation can mask the severity of cyanosis early on. However, once the ductus arteriosus closes—typically within the first day or two of life—oxygen levels in the bloodstream may decline significantly, leading to more pronounced cyanosis (8,9).

Infants may exhibit rapid breathing as a compensatory response to hypoxemia due to diminished pulmonary blood flow. Although a loud, harsh heart murmur is commonly associated with this defect, it might be absent during the initial days after birth. A sudden and dramatic drop in arterial oxygen saturation can also occur, known clinically as a “tet spell.” These episodes are typically caused by acute constriction of the right ventricular outflow tract, further limiting blood flow to the lungs (10).

During a tet spell, infants often become noticeably more cyanotic, with lips and skin turning a deep blue. These episodes are usually accompanied by intense irritability and hyperpnea. If the spell persists without prompt intervention, the child may become lethargic, unresponsive, or even lose consciousness due to critically low oxygen levels (8).

4. DIAGNOSIS

The prenatal diagnosis of Tetralogy of Fallot (TOF) using sonography remains challenging, mainly due to its reliance on the operator's skills and the lack of consensus regarding its sonographic indicators (11). Recent data suggests that only 42% of atrioventricular septal defects (AVSD) are detected through sonography (12). However, certain sonographic markers, such as thicker nuchal translucency, polyhydramnios, ventricular septal defect (VSD), and an enlarged aortic diameter, have been proposed as potential indicators for TOF (13). If there is a clinical suspicion of TOF, prenatal echocardiography and Doppler imaging should be conducted for a more accurate assessment.

While not absolutely essential in diagnosing TOF, chest X-rays have historically played a role. The classic view describes a "boot-shaped" heart due to the concavity of the pulmonary conus and the upward tilt of the cardiac apex. In TOF, the electrocardiogram (ECG) typically shows right axis deviation, with right ventricular hypertrophy (RVH) also appearing in the ECG, although the right axis deviation may not resolve as it would in a healthy newborn (14). As noted earlier, modalities such as multidetector computed tomography (MDCT) and magnetic resonance imaging (MRI) are emerging as useful alternatives. Additionally, cardiac catheterization remains valuable for assessing local vascular anatomy, which is critical for treatment planning (15).

5. MANAGEMENT

The development of pediatric cardiac surgery has been significantly shaped by advancements in the surgical treatment of Tetralogy of Fallot (TOF). In managing an acute hypercyanotic spell, several steps must be taken: providing high-flow oxygen (which may improve oxygen saturation once the shunt fraction decreases, and also helps reduce pulmonary vascular resistance), administering titrated sedation and analgesia (using morphine, fentanyl, or ketamine), increasing systemic vascular resistance (SVR) to mitigate the right-to-left shunt by using α-agonists like phenylephrine or noradrenaline, or vasopressin. If there is no improvement or in cases of severe hypoxia, intubation, mechanical ventilation, and neuromuscular blocking agents may be necessary. In cases of persistent infundibular spasm, esmolol or other β-blockers may be considered to slow the heart rate and improve diastolic filling (this should be done by experts only). If these measures fail, urgent surgical intervention may be needed, either as a palliative measure or to perform a complete repair (16).

For neonates and infants, cardiac catheter interventions can be used to improve pulmonary blood flow while waiting for the child to grow sufficiently for a full repair. This may involve pulmonary valvotomy or right ventricular outflow tract (RVOT) stenting, which can reduce RVOT obstruction and improve pulmonary blood flow (17). Additionally, stenting the patent ductus arteriosus (PDA) can decrease the need for intravenous prostaglandins in duct-dependent neonates. However, each of

these procedures carries its own set of risks.

Corrective surgery is typically performed before the age of 6 months as open intracardiac surgery under cardiopulmonary bypass conditions. This early intervention aims to reduce the pathophysiological changes associated with TOF, particularly the development of right ventricular hypertrophy (RVH) and subsequent fibrosis. The timing of surgery has been shown to reduce mortality and improve pulmonary vascular development. Many centers now perform primary repairs of TOF in neonates (18). The surgical repair involves patch closure of the ventricular septal defect (VSD), resecting muscle bundles in the RVOT, and reducing the severity of RVOT valve stenosis. A transannular patch may be necessary to enlarge the RVOT, but this leads to pulmonary regurgitation. Any associated conditions, such as atrial septal defects and PDA, are also addressed during surgery (16).

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