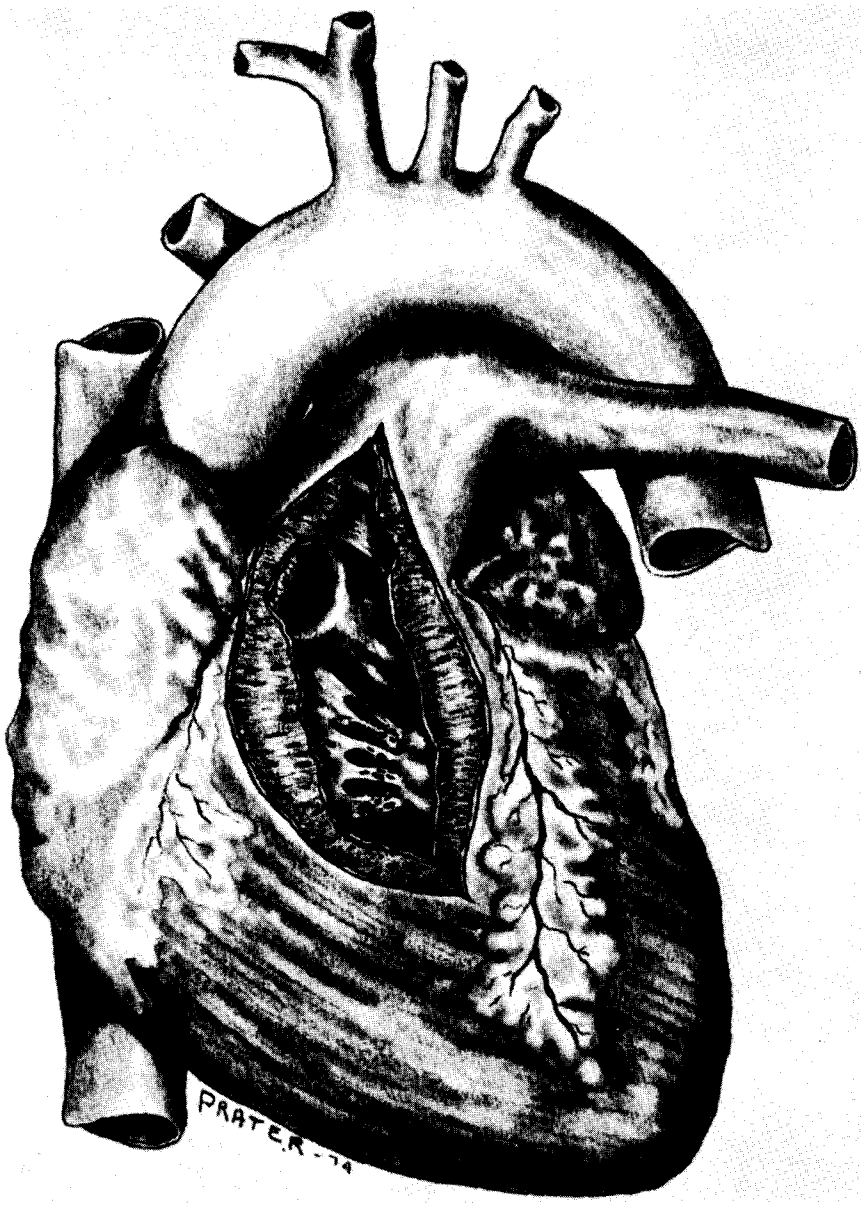

Tetralogy of Fallot

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In 1888, Fallot described a combination of cardiac malformations that includes ventricular septal defect, dextroposition of the aorta, pulmonic stenosis and right ventricular hypertrophy.[1] This combination is known as tetralogy of Fallot. The lesion is not, however, a clearly defined entity since there is considerable variation in the severity and site of the right ventricular outflow obstruction. There is also variation in the size of the ventricular septal defect and in the amount of dextroposition of the aorta.

Tetralogy is the result of a single embryologic error which gives rise to the complex of defects. In the embryo, the conus septum, which takes part in the division of the primative ventricular outflow tract (conus arteriosus) is located too far anteriorly. This causes the division to produce a small anterior outflow tract and a large posterior part. Since the displacement is more pronounced in the lower portion of the septum, the conus septum fails to meet with the interventricular septum and leaves a ventricular septal defect. This displacement also explains the dextropositioning of the aorta. Finally, the conus septum is so far removed that it cannot contribute to the formation of the tricuspid valve. The medial papillary muscle is absent and the valve malformed.[2]

The amount of pulmonary steriosis is quite variable. The right ventricular infundibulum is almost always stenotic. Although the pulmonary valve is often bicuspid, it offers significant resistance to flow in only 35 percent of the cases. The stenotic valve may be bicuspid or tri-leaflet or dome shaped with no clearly defined commissures.[3] In those patients with severe stenosis, the pulmonary blood flow is augmented by way of large bronchial arterial branches.

The ventricular septal defect is usually quite large and offers little resistance to flow between the ventricles. The defect may extend beyond the border of the membranous interventricular septum into the muscular portion of the ventricular septum.[4]

The amount of dextraposition of the aorta is also quite variable. The aorta appears to arise from the right ventricle and generally straddles the ventricular septal defect. The ascending aorta may be quite large and the main pulmonary artery hypoplastic or even atretic.[5]

Since the amount of right ventricular hypertrophy is dependent on the amount of work the right ventricle must do, the right ventricular wall may be thicker than that of the left ventricle. This hypertrophy may add to the right ventricular outflow obstruction.[3]

The pathologic physiology of the lesion is dependent upon the severity of the pulmonary stenosis and the size of the ventricular septal defect. Severe pulmonary stenosis leads to reduced pulmonary blood flow and oxygenation of blood. If pulmonary resistance is higher than systemic resistance, the shunt at the ventricular level is right to left. If the pulmonary resistance is lower than systemic resistance as in cases of mild obstruction to right ventricular outflow, the shunt will be left to right and the patient will present as a "pink tetralogy". Finally since the ventricular septal defect is a "relief valve" for the right ventricle in cases of severe pulmonary stenosis, the size of the ventricular septal defect influences the amount of work that the right ventricle must do and thus the amount of hypertrophy. If the defect is large and offers no resistance to flow, pressures are balanced and the right ventricular wall becomes approximately as thick as that of the left. If, on the other hand, the ventricular septal defect is small and offers considerable resistance to flow, the right ventricle will have to generate more pressure than the left and will become more hypertrophied.

The clinical appearance of patients with tetralogy is as varied as the pathology. Tetralogy of Fallot is the leading cause of cyanosis and clubbing of fingers in adults[6]. In the newborn, the defect does not generally create cyanosis until the ductus arteriosus closes. However, as the child develops and grows, the stenosis becomes relatively more significant. Cyanosis, observable only during periods of exertion at first, becomes noticeable at rest, usually within the first few years. Clubbing of fingers follows the appearance of cyanosis. Any change in pulmonary or systemic resistance may cause a cyanotic crises. Anesthesia that lowers peripheral vascular resistance can cause profound systemic hypoxia. Paroxysmal spells of cyanosis and syncope are characteristic of severe cases. The cause of these spells is not, as yet, clearly defined but may be due to sudden spasms of the right ventricular infundibulum.[7] These episodes may occur several times daily and may lead to death.

Another characteristic behavior of the child with tetralogy is the assumption of a knee-chest position when short of breath or tired. This "squatting" is presumed to either increase the systemic resistance or to lower right ventricular pressure by reducing venous

return. Whatever the cause, arterial oxygen saturation does go up with squatting.[8]

Peripheral desaturation leading to polycythemia and hematocrits of 55 percent are common. In cases of severe desaturation, red cell counts have been noted to be as high as 12 million per cubic millimeter with hematocrits over 75 percent. These extremely high concentrations of red cells lead to circulatory disturbances such as cerebral thrombosis and hemmorhage, epistaxis, hemoptysis and hematemesis.[9]

Treatment of tetralogy is both medical and surgical. There are many advocates of early total correction as well as those that advocate palliative surgery in patients up to 5 years of age. It has been demonstrated recently that only about 20 percent of the patients suffering from this defect will be adequately managed without surgery through their first four years.[10]

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