Definition

In anatomic terms, the malformation is characterized by four constant features: subpulmonary infundibular stenosis, ventricular septal defect, rightward deviation of the aortic valve with a biventricular origin of its leaflets, and right ventricular hypertrophy.

Although all hearts have comparable features, the malformation represents a morphological spectrum, with one morphological hallmark which unifies the overall entity: the anterocephalad deviation of the infundibular septum, the muscular structure separating the subaortic and pulmonary outlets, relative to the rest of the muscular septum.

Although unified in the sense of the septal malalignment, the patients with tetralogy of Fallot have significant variations in the precise anatomy of the ventricular septal defect, the nature of pulmonary infundibular and valvular stenosis, and the degree of aortic override, which account for the differences in hemodynamic consequences.

The ventricular septal defect is located in the membranous septum; it is subaortic and sometimes extends to the subpulmonic valve area. The ventricular septal defect is large, at least as large as the aortic valve, causing equalization of left and right ventricular pressures. The right ventricular outflow tract is hypoplastic and almost always obstructive. Occasionally at birth the right ventricular outflow tract shows no significant obstruction and results in the so-called “pink” tetralogy of Fallot. The pulmonary valve annulus is typically small in tetralogy of Fallot and the leaflets are deformed. In contrast to pulmonary valvular stenosis, the main pulmonary artery and the branch pulmonary arteries are small. There might be branch pulmonary artery stenosis as well as peripheral pulmonary artery stenosis in addition to the right ventricular outflow tract and pulmonary valvular stenosis. Bronchial arterial collaterals which connect to the peripheral pulmonary arteries are sometimes present. The main pulmonary artery is occasionally atretic and the pulmonary arteries are fed either by patent ductus arteriosus or collateral vessels. The aortic valve is typically large.

The most frequently associated anomalies are atrial septal defect or patent foramen ovale (40% of the patients), right aortic arch (25%), anomalous coronary arteries (10%), persistent left superior vena cava (8%), and major aortopulmonary collateral arteries with an extremely variable incidence, mostly correlated with the degree and duration of the cyanosis. Other associated malformations rarely reported are anomalous pulmonary venous connection, supravalvular mitral stenosis, cor triatriatum, atroventricular septal defect, multiple ventricular septal defects, restrictive ventricular septal defect, hypoplastic or absent infundibular septum, fibromuscular subaortic stenosis, valvular aortic stenosis, valvular aortic regurgitation, aortopulmonary window, patent ductus arteriosus, aortic coarctation, vascular ring, non-confluent pulmonary arteries, and anomalous origin of the left subclavian artery.
Surgical options

Palliative treatment: systemic-to-pulmonary artery shunt, consisting in a classical (nowadays almost abandoned) or modified Blalock-Taussig anastomosis with interposition of a PTFE tubular prosthesis between the subclavian artery and the ipsilateral pulmonary artery.

Repair: consists of two main steps: a) patch closure of the ventricular septal defect from a transatrial-transpulmonary or from transventricular approach with longitudinal ventriculotomy; b) right ventricular outflow tract reconstruction. This can be performed with different techniques:
- from the right atrium and/or from the pulmonary artery (transatrial-transpulmonary approach) or from the right ventricle (transventricular approach): incision with resection of the heavy trabeculations that bind the infundibular septum to the anterior right ventricular wall,
- infundibular (pericardial or synthetic) patch enlargement (transventricular approach),
- transannular (pericardial or synthetic, with or without monocusp) patch enlargement (transatrial-transpulmonary and transventricular approach) in case of inadequate size of the pulmonary valve annulus, measured with Hegar dilators, after pulmonary valvotomy performed from the pulmonary artery (transatrial-transpulmonary approach) or from the right ventricle (transventricular approach),
- pulmonary arteries enlargement in case of inadequate size (measured with Hegar dilators), with prolongation of the transannular patch in case of an inadequate left pulmonary artery, with a separate patch in case of inadequate right pulmonary artery,
- valved conduit, generally biologic, interposed between the right ventriculotomy and the pulmonary artery bifurcation, to bypass an anomalous coronary artery.

Pre-operative information

In addition to the demographic and clinical data, the following morphologic information is required in order to plan surgical repair:
- size and morphology of entire right ventricular outflow tract, the pulmonary valve and annulus, the main pulmonary artery and its branches (Figs. 3.7.1 and 3.7.2),
- the origin and course of the main coronary arteries (Fig. 3.7.3), particularly the presence of a major branch crossing the infundibulum,
- the end-diastolic volume of the left ventricle.

To better plan a palliative procedure, generally a modified Blalock-Taussig shunt, the following information is required:
- the side of the aortic arch (Fig. 3.7.2c),
- presence of an anomalous subclavian artery,
- size and morphology of the pulmonary arteries, and in particular the presence of stenotic origin of one of them (Figs. 3.7.2c and 3.7.4).

Fig. 3.7.1. Tetralogy of Fallot. CT scan, sagittal projection, showing severe obstruction of the right ventricular outflow tract, with practically pulmonary atresia (arrow); the size of the main pulmonary artery has been adequately maintained (diameter=9.9 mm) by the presence of functioning modified Blalock-Taussig shunt (LV left ventricle, RV right ventricle) (photograph courtesy of Dr. Mohamed Tawil)