Prenatal diagnosis of tetralogy of Fallot

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Abstract

Tetralogy of Fallot makes up about 10% of all congenital heart defects and belongs to infundibular defects with anomalous origin of arteries. Its classic form is usually not life-threatening to an infant. It is characterized by VSD (ventricular septal defect), overriding aorta, pulmonary valve stenosis. Postoperative mortality ranges from 2-5%. Prenatal diagnosis of complex heart defect enables planning perinatological care. This article presents the case of a fetal diagnosis of the heart defect, diagnostic and therapeutic procedures.

Key words: tetralogy of Fallot (ToF), right ventricular outflow tract obstruction (RVOTO)

Case presentation

The primigravida without any history of obstetric complications was admitted to the Department of Perinatology and Gynecology because of a suspicion of fetal Fallot’s syndrome. At 25 week of gestation, an abnormal four-chamber view was observed. There was a ventricular septal defect (VSD) of 2 mm visible. Aorta valve of 6 mm was shifted over the VSD. The blood flow velocity through the aortic valve was normal. There was a narrow pulmonary trunk (PA) (of 3 mm in the diameter), with a blood flow velocity up to 160 cm/s and DA of 2.8 mm in the diameter. Abnormal three-vessel view in the mediastinum was found – aorta was shifted to the right side, right-sided aortic arch was noted. Fallot’s syndrome with right ventricular outflow tract obstruction (RVOTO) was diagnosed. Karyotyping was offered but the patient did not accept it.

The next examination revealed an increased blood flow velocity through the aortic valve reaching 112 cm/s. Holosystolic aortic valve regurgitation of 200 cm/s was also observed. The blood flow velocity through a narrow pulmonary trunk was high up to 170-200 cm/s.

Fig. 1. Abnormal blood flow through the TV

Fig. 2. Increased blood flow velocity through the pulmonary trunk

The patient went into a spontaneous delivery at 39 weeks’ gestation. The infant was admitted to the Department of Neonatology, where echocardiography confirmed a subaortic VSD (ca. 0.4 cm). Aorta was shifted to the right side. Flow in the descending Ao was measured - MFV = 0.7 m/sec as well as in the abdominal Ao - MFV = 0.23 m/s. A turbulent flow in PA was observed (MFV = 3 m/s) and the diameter of the DA was measured (0.15 cm) with L-P flow. On the first day of life, the newborn was transferred to the Department of Cardiology and Nephrology at Karol Jonscher’s University Hospital in Poznań. After a few days of observation the

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initial diagnosis was confirmed. The infant was discharged from the hospital and outpatient cardiology consultations was recommended. Surgical treatment was deferred until the infant was six months of age.

Fig. 3. Aortic valve of 6 mm shifted over the ventricular septal defect

Fig. 4. Three vessels view in the mediastinum shows an arrow hipoplastic pulmonary trunk

Discussion

Tetralogy of Fallot is the most common cyanotic congenital heart defect, representing about 10% of all congenital heart defects [1, 2]. It occurs with a frequency of 3.6 per 10,000 live births [3].

The anatomical scope of this defect is large, and therefore its clinical course varies. The view of the four-chamber heart may be normal, because the VSD is in the subaortic area which is located at the front in relation to the section of four heart chambers. Such location results in shifting the aorta over the right ventricle (dextroposition of the aorta), which results in the view “of overriding aorta”.

The axis of the heart is abnormal, the angle exceeds 45°. Morphologically, the Fallot syndrome is characterized by shifting the infundibular septum forward, up and to the left. This shift results in a large ventricular septal defect and narrowing of the right ventricular outflow tract. In the mediastinum image (the image of the four heart chambers), the pulmonary artery is located in the front and is narrower than the aorta, a small thymus is visible.

Diagnosis of the TOF is facilitated by Doppler echocardiography. Usually there is a variable degree of blood flow acceleration in the pulmonary artery above 1 m/s. When the narrowing of the right ventricular outflow tract is significant, pulmonary arteries may become hypoplastic due to the very small volume of the blood flowing. In such cases, it may be a ductus-dependent defect, which means that the life of an infant depends on maintaining patency at the level of the ductus arteriosus. Other defects of the vascular system include: persistent left superior vena cava draining into the coronary sinus, right-sided aortic arch, anomalous origin of the subclavian artery [4].

According to Palladini et al. [6], in approximately 45% of fetuses with ToF, non-cardiac defects are diagnosed: umbilical hernia, pentalogy of Cantrell, VACTERL or CHARGE syndromes. Moreover, in 25% Fallot syndrome can coexist with chromosomal aberrations such as: Down syndrome, trisomy 18, triploid syndrome and microdeletion of chromosome 22. For this reason, the evaluation of karyotype should be considered [7, 8].

Different types of TOF defect may be distinguished. TOF with right ventricular outflow tract obstruction includes four-chamber view close to normal. VSD has a location typical for the TOF, but there is no connection between the right ventricle and the pulmonary trunk. Pulmonary trunk and branches are hypoplastic, the right ventricle outflow tract is obstructed, and the pulmonary vascular bed is filled by the MAPCAs (main aorto-pulmonary collaterals), bronchial vessels, rarely by the ductus arteriosus [8]. In the mediastinum three incorrectly located vessels may be visible with a narrow vessel located at the front. The diameter of the pulmonary artery does not exceed 2-3 mm. This defect must be differentiated from the common atrioventricular canal.

Another form of TOF is an anomaly with absent pulmonary valve. This is a rare heart defect. Heart anatomy is similar to that in ToF (movement of infundibulum and the typical VSD location). The pulmonary valve ring is hypoplastic. There may be rudimentary valve leaflets or they may not occur at all. The lack of ductus arteriosus and a large septal defect are diagnosed. In this defect, the blood flowing from the right ventricle to the pulmo-
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nary artery, cannot flow into the descending aorta because of the lack of the ductus arteriosus, and pulmonary vascular resistance is high. This causes broadening of the two branches of the pulmonary arteries. They can be seen as broad, vibrant, hypoechogenic formations in the mediastinum.

Due to the fact that blood cannot flow freely in the high-resistance pulmonary circulation, it retracts into the right ventricle, leading to the destruction of the pulmonary valve leaflets. The blood which retracts from the pulmonary trunk to the right ventricle increases the volume in the right ventricle, which leads to mitral regurgitation, right atrial enlargement, increase of venous pressure and consequently, to heart failure, including generalized oedema [9].

In the prenatal ultrasound an abnormal four-chamber view, often enlarged and abnormal right ventricle contractility may be seen. The branches of pulmonary arteries are enlarged and their image is often considered to be the image of an enlarged left atrium. In this case, from the middle period of gestation, the pulmonary arteries may have a diameter of 10 mm (normally, at this time at this time its diameter does not exceed 3 mm), approximately. Colour and pulsed Doppler show systolic and diastolic flow through the pulmonary trunk.

Surgical correction can usually be deferred and planned depending on the clinical condition of the infant, its development and anatomical conditions ascertained after birth. In infants with ductus-dependents pulmonary blood flow, prostaglandin E1 is administered by a constant intravenous infusion. This treatment is continued until the completion of diagnostic tests, interventional or surgical procedure. The diagnosis of ToF is a priori indication for surgery or cardiac interventions. The choice of surgical treatment and its time is debatable. If the diagnosis of the defect is made in a symptomatic newborn during the first three months of life, most authors support the two-stage treatment and the implementation of Blalock-Taussig shunt or baloon valvuloplasty for pulmonary valve at the first stage. The advantage of Blalock-Taussig shunt is that it is being performed outside the pericardium, which prevents adhesion and facilitates the closure during defect correction. An alternative method is a balloon valvuloplasty for pulmonary valve stenosis. Effective balloon valvuloplasty for pulmonary valve stimulates the development of pulmonary artery ring and its branches, and significantly improves oxygen saturation.

Postoperative mortality ranges from 2 to 5%. The prognosis for ToF correction is considered good. According to many reports, about 95% of patients survive until 25 years after the surgery, and 85% of patients survive until more than 30 years after the surgery. In the years 1981-2000, in the IPCZD Department of Cardiac Surgery, a total of 780 ToF surgeries were performed, with a mortality rate of 6.8%. In 1996-2000, a total of 179 ToF surgeries were performed, with death occurring in six cases, which constitutes 3.35% [11].

From a surgical point of view, the most important factor is to assess the morphology of the right ventricular outflow tract, pulmonary valve and the degree of development of pulmonary trunk and arteries. For this purpose, the ultrasound testing is performed using a new 4D diagnostics with the B-flow and STIC imaging technique. New imaging techniques may be helpful in determining the source of pulmonary vasculature and the complete picture of the development of pulmonary branches as well as accurate prenatal diagnosis of complex heart defects [12].

References


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