DIAGNOSTIC STAGES FOR FALLOT DISEASE IN NEWBORNS AND INFANTS

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Abstract

Introduction-Purpose of study. Fallot disease is a cyanotic heart malformation witch if diagnosed in an early stage can be optimally corrected trough surgery leading to a normal life style for the patient. Quantifying the clinical situation, systolic murmurs, low oxygenation, hemodynamic instability, fatigue, peripheral cyanosisand phenotypical dismorphism, need an: x-ray, echocardiography, cardiac MRI.Clinical signs, laboratory and imaging investigations establish the diagnosis and suggest the correct treatment. *Material and method*. The study was carried out using 16 newborn and infant diagnosed with Fallot in newborn Clinics in Timisoara, in 2015-2016. The diagnosis was suggested by the clinical evolution of the subjects and confirmed by echocardiography in most cases, some in utero. *Results.* Statistically most of the cases were diagnosed as newborns, when echocardiography was used to investigate a systolic murmur. The in utero diagnosis was useful in suggesting a place of birth in close proximity to a cardiovascular center in the cases of extreme Fallot disease. The genetic syndrome association with Fallot disease was present in 3 of the cases. *Conclusions.* 1. The neonatal screening is important in cardiac malformation cases witch require surgery. 2. The morphological variability translates in the clinical state of the patient and emergency degree, so the in utero diagnosis is very significant. 3. The morphological complexity and severity of the case was often associated with other lesions in the case of a genetic syndrome.

Key words: Tetralogy Fallot, cardio-vascular signs

Introduction - The purpose of the paper

Tetralogy of Fallot (TOF) is a cyanotic congenital heart malformation, is considered the prototype of cyanotic congenital heart malformations. The four classic morphological malformations are the following: ventricular septal defect, right-sided aortic arch, and pulmonary artery stenosis and right ventricular hypertrophy.[1,2]

In addition TOF may present other anatomical anomalies including: an atrioventricular septal defect, total or partial abnormal pulmonary venous return, coronary artery abnormalities, a patent foramen ovale or atrial septal defect, in which case the syndrome is sometimes called a pentalogy of Fallot.[3-6]

A severe variant of TOF is the type with complete obstruction (atresia) of the right ventricular outflow tract. In these individuals, blood flows from the right ventricle to the left where it is pumped only through the aorta. The lungs are perfused via extensive collaterals from the systemic arteries, and sometimes also via the ductus arteriosus.[3-5,7,9-12]

In the case of the infant who escapes the neonatal filter without being diagnosed, we can also observe sucking fatigue, varying degrees of shortness of breath, weight curve slowly upward or stagnant, syncopal episodes, fainting, repeatedear, nose and throat infections, brain abscess.[7,8,10]

Clinical signs and symptoms vary depending on the age of the patient at diagnosis and are nonspecific, but the presence of a systolic murmur raises the suspicion of congenital heart diseases. During the newborn stage different degrees of systolic murmurs and o2 desaturation can appear if the newborn is agitated or nursing, also hemodynamic instability , peripheral cyanosis , phenotypic dysmorphism absence of clinical symptoms or only present a systolic murmur can, prematurity or low weight for gestational age can be observed.[1,8,12]

Quantifying the clinical situation, systolic murmurs, low oxygenation, hemodynamic instability, fatigue, peripheral cyanosis and phenotypical dysmorphism, need an: x-ray, echocardiography, cardiac MRI. Clinical signs, laboratory and imaging investigations establish the diagnosis and suggest the correct treatment.[13-18]

Material and method

The study was carried out using 16 newborn and infant diagnosed with Fallot in newborn Clinics in Timisoara, in 2015-2016. The number of patients included in the extended retrospective doctoral study is 80 patients with TOF but the criteria for the present paper limited the number.

Patients diagnosed postpartum showed intense specific systolic murmur (pulmonary stenosis), and the echocardiography confirmed the diagnosis. We can say that 7 cases of typical Fallot were diagnosed in Maternity, post echocardiographic review of systolic murmurs. Patients were hemodynamically stable with varying degrees of cyanosis or sucking fatigue, with systolic murmurs of different intensity, they came from healthy parents and the pregnancies were monitored. Of the 7 cases, 4 were solved surgically, surgical correction being performed per primam between the ages of 4 months and 1.6 years, with good postoperative evolution without major problems and age-appropriate social insertion.

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We diagnosed echocardiographic a 5 months old infant with TOF and agenesis of eyeballs with systolic murmur, undiagnosed completely until this age, when the first hypoxic crisis occurred - in the context of a respiratory infection. Other associated malformations, maternal intrauterine infection or association with a genetic syndrome were excluded through imaging and laboratory means. Currently is 11 months old, it is stable and will soon benefit from surgery per primam.

We recruited two cases of tetralogy of Fallot with pulmonary atresia diagnosed in maternity with cyanotic heart malformation incompletely defined. The cases had loud systolic murmur, cyanosis, hypoxic seizures, and at 2 and 6 months were correctly diagnosed through imaging (echocardiography, Rx, angioCT), with Tetralogy of Fallot with pulmonary atresia (PA) and major aortopulmonary collateral arteries (or MAPCAs). These cases showed extensive collaterals and are suitable for surgery with aortal -pulmonary collateral focus and total correction.

TOF with PA is considered the extreme morphopatological variant. Pulmonary atresia may be limited to the valve (pulmonary atresia membranous) or infundibular subvalvular (muscular pulmonary atresia). [19-23]The lack of ante grade pulmonary blood flow in utero leads to a range of morphologic findings in the pulmonary artery vasculature such as, confluent true pulmonary arteries of variable sizes if the ductus arteriosus (DA) is present. Without flow through the DA, MAPCAs, fetal vessels derived from the splanchnic vascular plexus, may persist after birth. These vessels connect the systemic and pulmonary arterial vasculature, thereby supplying pulmonary blood flow. MAPCAs are tortuous vessels that arise directly from the aorta or its branches and vary in number and origin, follow routes to reach central, lobar, and segmental pulmonary arteries, and have variable areas and locations of stenosis. The morphology of the pulmonary vasculature and MAPCAs plays a critical role in determining management decision. [19,20]

We have included 3 cases of Fallot diagnosed intrauterine were fetal ultrasound raced a high suspicion of Fallot, these cases were confirmed by echocardiography postpartum, in the presence of systolic murmur and peripheral cyanosis. Because of hemodynamic stability, mild and rare hypoxic crisis, light/medium pulmonary stenosis, benefited from achieving total correction, optimally between 6 months and 1 year with very good postoperative adjustment.

We observed the association between DiGeorge syndrome and Fallot in 3 cases, in one case there was an antenatal diagnosis of congenital heart malformation. Patients had pulmonary atresia and multiple stenosis, in series of pulmonary branches. They presented systolic murmur, intense cyanosis, hemodynamic instability and PG therapy was established to keep the ductus arteriosus permeable, in order to find solutions for urgent surgical treatment. One patient received systemic-pulmonary shunt at 3 months old and then 3 pasty with stenting of pulmonary branches post cardiac catheterization approximately every 6 months / years, with complicated evolution and prognosis in the context of genetic syndrome and stenosis of multiple branches. The second patient with TOF associated genetic syndrome (who was diagnosed antepartum), presented a giant aneurysm of pulmonary trunk, pulmonary regurgitation grade III / IV. It has received a total correction at age 3 months. The third case is in the phase of investigation the genetic syndrome by karyotype and molecular techniques, is phenotypically classified in DiGeorge syndrome. It has benefited from prostaglandine (PG) therapy, total correction for Fallot the age of 4 months, later two pulmonary plasty post cardiac catheterization for stenosis of pulmonary branches. It is currently stable with good performance.[24-26]

Results and Discussions

Statistically most of the patients 56% have been diagnosed in maternity after the evaluation of systolic murmur. (Fig.1)

A total of 68% of the patients in study have been diagnosed with classical TOF, 3 cases (18%) with TOF with DiGeorge genetic syndrome and 2 cases (12.5%) with TOF with PA. (Fig.2)

11 patients (68%) from the study group had surgery, 10 patients (62%) benefitted of total correction per primam. 1 patient was initially stabilized by shunt systemic- pulmonary at age 3 months, and then received total correction surgery at 1 year old, as well as two plasty with stenting of pulmonary branches.

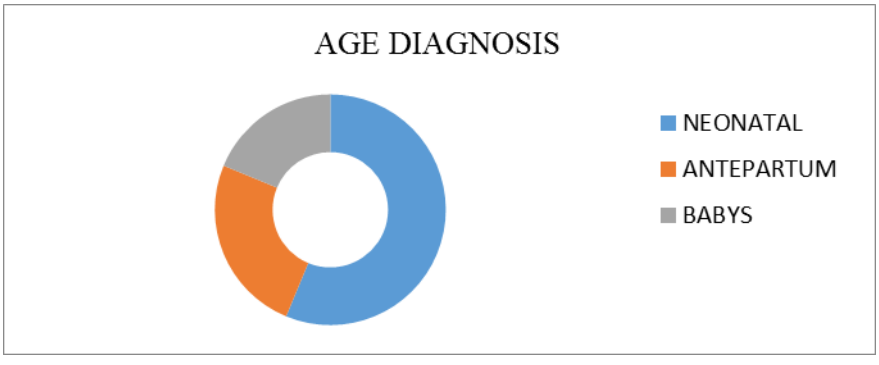

Fig. 1. Age of patients at the moment of diagnosis.

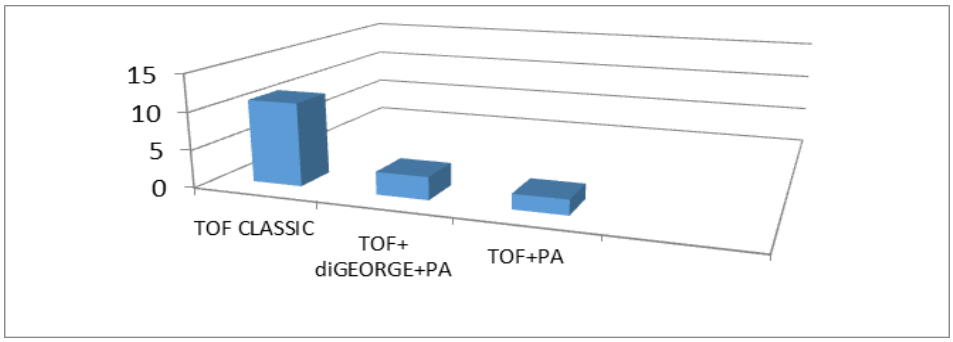

Fig. 2. The number of TOF (classic and extreme) and TOF with genetic syndrome association.

We could not perform surgical correction per premium due hemodynamic instability, severe hypoxic crises and malformation context.[2,28,29]

There were no deaths registered in the group of patients that received surgery or in the group that awaits correction surgery. (Fig.3)

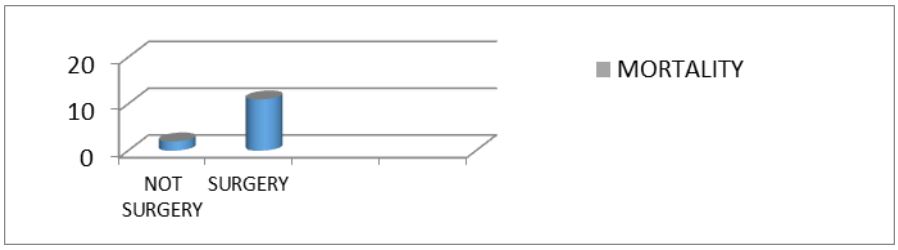

Fig. 3. The number of patients with and without surgical correction.

The in utero diagnosis was useful in suggesting a place of birth in close proximity to a cardiovascular center in the cases of the newborn with the extreme type of Fallot disease.

The prognosis is good for classic form TOF patients, because they have been diagnosed on time and have received or will receive timely total correction. In the case of the patient with TOF with PA the prognosis is reserved due to the many collateral arteries and surgery limitations. Patients with TOF and DiGeorge syndrome have a high-risk of developing complications and a reserved prognosis. (Fig.4)[2,30]

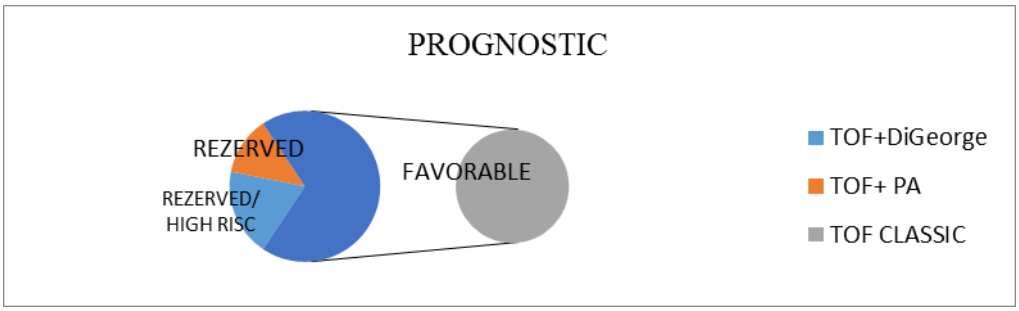

Fig. 4. Prognostic of TOF (classic, extreme, TOF with genetic syndromes).

Conclusions

1. The neonatal screening is very important in the case of cardiac malformations with surgical indication.

2. Echocardiography established the correct diagnosis for infants with cyanosis and systolic murmur and indicated the optimal surgery needed to correct the problem.

3. The morphological variability translates in the clinical state of the patient and emergency degree, so the in utero diagnosis is very significant.

4. The morphological complexity and severity of the case was often associated with other lesions in the case of a genetic syndrome.

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