INTRODUCTION

The Ebstein anomaly (EA) is a congenital heart defect in which the tricuspid valve (TV) has displazic leaflets with altered structure and/or position. In the normal heart there are three TV leaflets: anterosuperior (or anterior), inferior (or posterior) and medial (or septal). Every normal TV leaflet consists of a peripheral zone to which the chordae tendineae inserts, a basal zone of atrioventricular attachment, and a free zone between these two. The anterior leaflet is the largest of the three. The septal leaflet is smaller than the other two and has an apical insertion compared with the anterior leaflet of the mitral valve.

In EA, one or more tricuspid leaflets are displaced. The septal or/posterior leaflets of the TV are displaced from the tricuspid annulus toward the right ventricular apex (there is more than 1 cm or 0.8 mm/m² body surface area, between the mitral plane and the displaced TV). The anterior leaflet is large and sail-like with abnormally numbered and placed chordal attachments. As a result, the valve orifice is displaced downward into the right ventricle (RV). These changes lead to the atrialization of a part of the RV. The inlet portion of the RV is functionally integrated within the right atrium (RA), which becomes the atrialized RV, while the others, trabecular and outlet portions, constitute the functional RV. The RV area between the true tricuspid annulus and the displaced attachment of the septal and posterior leaflets is thinned and dilated and the remaining right ventricular cavity is small and has the characteristics of the RV wall. Sometimes the interventricular septum has a...
paradoxical motion which alters the left ventricular (LV) geometry and function. Atrialized RV volume/total RV volume ratio more than 30% indicates severe disease with poor prognosis. Commonly, the tricuspid regurgitation is severe with low velocity on continuous wave Doppler. An interatrial communication is also frequently present.

CASE REPORT

We want to report the case of a young man, 29 years old, from city environment, admitted in our Clinic in May 2007, with the following complaints: dyspnea at medium effort, asthenia and peripheral cyanosis and a systolic murmur which covers the entire precordial area. Working and living environment were normal.

Clinical examination revealed a hypostenic patient (height = 174 cm, weight = 60 kg) with normal pulmonary sounds on auscultation, regular cardiac rhythm, 110 beats/min, blood pressure 90/60 mmHg, ejection systolic murmur of maximum intensity grade V/VI which covered the entire precordial area and clubbing.

Biological data were within normal limits.

The electrocardiography showed a regular sinus rhythm, intermediary QRS axis, 80 beats/min and depressed ST segment in DI, DIII, aVF.

Chest X-ray demonstrated a global heart enlargement.

Transthoracic echocardiographic exam showed normal size and function of the LV and left atrium (LA), normal LV kinesia and dilated right chambers. The mitral and the aortic valves were normal and the valvular flows were within normal limits. The interventricular septum had a paradoxical movement. (Fig. 1)

In the apical four chamber view, the apical displacement of the tricuspid septal leaflet and the long anterior leaflet, with the gear attached to the free right ventricle wall, can be viewed in Figure 2. The displacement of the septal tricuspid leaflet was measured as the distance from the anatomic annulus to the distal attachment of the septal leaflet and was 3.2 cm/m² body surface area. (Fig. 1) The posterior leaflet was short and small. The septal tricuspid leaflet had a small echodense mass attached to it, which swung between the RA and the RV. (Figs. 3, 4)

The atrialized RV volume was 187 ml and total RV volume 274 ml. (Fig. 5a,b) The difference represented the functional RV, which measured 135 ml.

There was tricuspid regurgitation grade IV with Pmax 20 mmHg and evaluated systolic pulmonary arterial pressure (SPAP) 25 mmHg. (Fig. 6)

Transesophageal echocardiography confirmed the displacement of the posterior and septal leaflets, the
tricuspid regurgitation grade IV and also the flail of septal leaflet. (Fig. 7a,b) This last image raised the suspicion of a ruptured septal tricuspid leaflet.

Figure 4. Apical 4 chamber view. Note the atrial mass at the level of septal tricuspid valve (arrow). LV – left ventricle, RV – right ventricle, LA – left atrium, RA – right atrium.

Figure 5. Apical 4 chamber view. The measurement of total end-diastolic right ventricle volume, 274 ml (a) and the atrialized right ventricle volume, 187 ml (b). The difference represents the functional right ventricle volume: 274 – 187 = 87 ml. LV – left ventricle, LA – left atrium, RA – right atrium.

Figure 6. Continuous-wave Doppler at the level of the tricuspid valve, from apical 4 chamber view, shows the low velocity severe tricuspid regurgitation (TR).

Figure 7. Transoesophageal echocardiography: two different sections show the tricuspid septal leaflet (s) with flail and rupture suspicion (arrow). The anterior tricuspid leaflet (a) is large and the posterior tricuspid leaflet is small (p). RA – right atrium, RV – right ventricle.

TEE also showed an atrial septal defect (ASD) of sinus venosus type near the superior vena cava (SVC) entrance in the RA, with a diameter of 2.6 cm and a left-to-right shunt. (Fig. 8) The anterior tricuspid leaflet was long with the gear attached to the RV wall.

The positive diagnosis established after these evaluations was:
- Ebstein anomaly with severe tricuspid regurgitation;
- Septal tricuspid leaflet rupture;
- Sinus venosus atrial septal defect with left-to-right shunt;
- Class III/IV NYHA cardiac failure.
Figure 8. Transoesophageal echocardiography, section at 62°, shows the atrial septal defect type sinus venosus near the superior vena cava (SVC) entrance in right atrium (RA), with a diameter of 2.6 cm and a left-to-right shunt on color Doppler (arrow). LA – left atrium.

We considered the surgery as the unique therapeutic solution for this patient.8-10

The reasons for surgical indication in our patient were:
- Functional class III NYHA symptoms;
- Progressive decline in exercise tolerance, fatigue;
- Severe cardiomegaly;
- Atrialized RV volume/total RV volume ratio = 68%.

The goals of the surgical intervention were as follows: to increase pulmonary blood flow, to minimize tricuspid insufficiency, to reduce or eliminate left to right shunt, to optimize right ventricular function.8-10

The operation was performed using cardiopulmonary bypass, bicalv cannulation, moderate systemic hypothermia, and cardioplegia for myocardial arrest.

The intraoperatory findings were consistent with the echocardiographic description. The RA was extremely enlarged, but that represented in fact the atrialized RV. After the RA was opened, a large septal atrial defect of 2.5/3.5 cm, near SVC was exposed.

The septal leaflet was found to be rudimentary, ruptured and downward displaced into the RV, the posterior leaflet was hipoplasic and the anterior one had normal dimensions and positioning as it can be observed in Figure 9. The Danielson repair was performed.8 It consisted of a plication of the atrialized RV in a horizontal plane, posterior tricuspid annuloplasty, closure of the ASD, and RA reduction. (Fig. 10)

The plication of the atrialized RV brought the base of the posterior leaflet into the plane of the anterior leaflet. This repair essentially created a competent monocuspid valve from the large anterior leaflet, which coapts to the free edge of the relocated posterior leaflet and to the septal leaflet.

The postoperative evolution of the patient was good and he was discharged from the hospital with the following medical treatment: β and α blockers, diuretics and anticoagulants.

The prognosis of the patient is considered to be good. Long-term follow-up after surgery shows continuing improvement, even though the patient had associated defects and severe symptoms, a cardiothoracic ratio greater than 0.65, and class III NYHA, which are all predictors of increased mortality.

The follow-up will be done after the first six months, then annually, with clinical, echocardiographic and Holter monitoring reevaluation according to the European Guidelines recommendations.11

DISCUSSION

Slightly less than 1% of all newborn infants have congenital heart disease. Eight defects are more common than all others and make up to 80% of all congenital heart diseases, whereas the remaining 20% consist of many independently infrequent conditions or combinations of several defects. EA is a part of those 20%.
Wilhelm Ebstein described the first case of EA in a young man, at the autopsy. Later, in 1950, the first diagnosis of EA was made in a living patient. In the 1960s EA was believed to be a very rare disease (< 0.3%). The diagnosis at that time was a sum of clinical, electrocardiographical and sometimes angiographical data. Since the echocardiography has become a routine investigation the number of patients with this diagnosis has increased, however not so much, and today it still is a relatively uncommon diagnosis in children and adults with congenital heart diseases.  

After a three year study that included 875 fetal echocardiograms, Macedo et al found 4.2% cases of congenital heart disease. From the total 875 echocardiograms 5 of them presented malformations of the tricuspid valve. EA represented approximately 45-60% of all TV disease. The reported prevalence was 0.3-0.5% among congenital heart defects. 

Clinical diagnosis of EA in adults is difficult, because it actually is a differential diagnosis of a systolic murmur in a patient with right heart failure, that includes: ventricular septal defect, aortic stenosis, mitral regurgitation, mitral valve prolapse, and pulmonary stenosis. The echocardiography made the diagnosis easier: it showed a normal size and movement of the left ventricle, normal mitral, aortic and pulmonary leaflets and also no interventricular septal defect. It showed a severe, but low velocity tricuspid regurgitation and the displacement of the tricuspid leaflets. The differential diagnosis can also refer to different anatomic and hemodynamic variables of EA: other forms of congenital tricuspid regurgitation (unguarded tricuspid valve, pure tricuspid valve dysplasia), imperforate tricuspid orifice or pulmonary atresia and intact interventricular septum.

About 50% of individuals with EA have an associated shunt between the left and right atrium, either an ASD or a patent foramen ovale.

Echocardiography can define the specific abnormalities of the TV leaflets; the size and the function of the RV can also be assessed. Coexisting cardiac lesions can also be identified. Color flow Doppler allows the assessment of the tricuspid regurgitation and the degree of shunting at the atrial level. A well-documented correlation exists between the echocardiographic findings and those during surgery.

Two systems that classify disease severity have been developed, based on echocardiographic findings. The classification of Carpentier is based on the TV morphology and RV volume, while the classification of Celermajer is based on right cardiac chamber size.

Carpentier classification: 
- Type A: RV volume is adequate;
- Type B: large atrialized segment of the RV and mobile anterior leaflet;
- Type C: restricted movement of the anterior leaflet, which may cause infundibulum obstruction;
- Type D: near-complete atrialization of the RV (Uhl's syndrome).

Celermajer et al described an echocardiographic grading score for neonates with Ebstein Anomaly, GOSE (Glasgow Outcome Score Extended) Score Grade 1–4. For this score, the ratio of the combined area of the RA and atrialized RV and that of the functional RV and left heart in a four-chamber view at end-diastole was calculated (ratio < 0.5, grade 1; ratio 0.5–0.99, grade 2; ratio 1.0–1.49, grade 3; and ratio > 1.5, grade 4).

More recently, real-time 3-dimensional trans-thoracic echocardiography has provided a more comprehensive assessment of the anatomic and morphologic features in patients with EA, regarding the apical displacement of the septal tricuspid valve leaflet: at least 8mm/2mm displacement of the septal leaflet from the anterior mitral valve insertion.

Valvular dysplasia present in EA is defined as valvular thickening, nodularity and irregularity. EA is associated with RV dilatation in approximately two-thirds of the cases. Dilatation may be massive and often involves not only the atrialized RV (proximal to the TV) but also the functional RV (distal to the TV), including the right ventricular infundibulum. Sometimes the dilatation is so important, that the interventricular septum bulges in the LV. For further definition of the pathogenesis of dilatation of the functional RV (distal chamber), a morphometric histopathologic study was performed on 10 hearts with EA and 10 normal age-matched control hearts. In the group with EA, five hearts exhibited dilated ventricles and five did not. The study demonstrated that dilatation of the RV in EA was associated not only with thinning of the wall but also with an absolute decrease in the number of myocardial fibers counted through the thickness of the wall from endocardium to epicardium. Marked enlargement of the RA and atrialized RV is considered to be present if the combined area of the RA and atrialized RV is larger than the combined area of the functional RV, LA and LV, measured in the apical four chamber view at end-diastole.

The site and degree of tricuspid valve regurgitation and the feasibility of valve repair are also assessed by echocardiography. The downward displacement of the septal
tricuspid valve leaflet is associated with discontinuity of the central fibrous body and septal atrioventricular ring, thus creating a potential substrate for accessory atrioventricular pathways and ventricular pre-excitation, exposing the patient at the risk of sudden death. At this moment, the patients with symptomatic arrhythmias have an indication for electrophysiological investigations and radiofrequency ablation in selected cases. In a study made at the Mayo Clinic on 52 patients with EA, 3.7% needed permanent pacing for atrio-ventricular block (most of them) or sinus node dysfunction. Angiography has demonstrated that a significant number of patients with EA have morphological and functional abnormalities of the LV, which may be explained by increased fibrosis in the LV wall and ventricular septum as demonstrated by histological studies. Daliento et al analysed the left ventricle in 26 patients with EA and showed that 12 had enlarged LV and 8 of them also had depressed LV systolic function. Moreover, they observed that functional alteration of LV appeared only when LV was dilated.

The indication for surgery is made by the patient’s symptomatology: progressive deterioration of clinical status, with evident cyanosis, right cardiac failure (class III/IV NYHA), paradoxical emboli, ventricular tachyarrhythmias. Patients have good long-term survival and functional outcomes after undergoing surgery for EA. There are different techniques which repair the tricuspid valvular apparatus with or without internal plication of the atrialized RV.

Ebstein repair has good functional outcomes despite residual tricuspid regurgitation, likely because of the reduction in right ventricular volume loading. Adult patients did not demonstrate the same durability of valve repair like children and frequently required tricuspid valve replacement. Intraoperative radiofrequency ablation represents an important adjunctive treatment for intractable arrhythmias, which may now represent relative indications for operative intervention, but this was not the problem with our patient.

CONCLUSION

The preoperative diagnosis and postoperative follow up in patients with EA make a very important issue, and the gold standard investigation for this is echocardiography, which provides information about TV morphology and about right and left ventricle systolic and diastolic functions.

EA is a well defined congenital anomaly, but its therapeutic strategy is very complex. The anatomical alterations, their functional significance and the therapeutical options must be evaluated individually in every patient with EA.

REFERENCES

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