EBSTEIN’S ANOMALY

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EBSTEIN’S ANOMALY (Abstract): Ebstein's anomaly is a rare congenital heart disease, accounting for less than 1% of all congenital heart diseases, characterized by a wide clinical, electrocardiographic, echocardiographic, anatomic and prognostic polymorphism. The disease can be fatal since birth or may remain asymptomatic until adulthood, sometimes being associated with septal defects, transposition of great vessels, preexcitation syndromes, or left ventricular noncompaction. The genetic changes underlying this syndrome are not fully known, but in the cases associating left ventricular nonompaction a mutation in MYH7 gene encoding the β-myosin heavy chain was recently detected. The authors present 2 cases of Ebstein's anomaly with different onset and course and discuss the current clinical, electrocardiographic and echocardiographic criteria used for prognostic stratification of Ebstein disease in relation to international literature. Keywords: EBSTEIN’S ANOMALY, TRICUSPID VALVE, ELECTROCARDIOGRAPHY, ECHOCARDIOGRAPHY.

Ebstein’s anomaly is a rare congenital cardiac malformation characterized by a combination of pathological anatomical and functional changes of the tricuspid valve, right ventricle and atrium. The diagnosed cases account for less than 1% of all congenital cardiac malformations, the prevalence being approximately 1 in 20,000 live births (1-4). The malformative complex affecting the tricuspid valve and right ventricle is characterized by many variations and degrees of severity, conferring a significant clinical electrocardiographic, echocardiographic and evolutive polymorphism to the disease. The main lesion is represented by the apical displacement of the septal and posterior tricuspid valve leaflets by over 8 mm/m² body surface area. Functionally, the malformation causes atrialization of part of the right ventricle, and different degrees of tricuspid regurgitation, sometimes associated with stenosis. The disease can be associated with other cardiac malformations, most commonly with interatrial septal defect or patent foramen ovale (80-94% of patients) and rhythm and conduction disorders, due to the discontinuity of intracardiac excito-conductor system (3-6). The authors discuss the peculiarities encountered in two cases of Ebstein's anomaly diagnosed in adults and compare them with the literature data.
CASES PRESENTATION

Case 1. A 51-year-old man was admitted for cardiovascular symptoms that occurred about 2 years earlier and gradually worsened, including NYHA class II exertional dyspnea, atypical chest pain without specific radiation, enhanced by mental strain and fatigue. History did not reveal other cases of congenital heart disease in consanguineous relatives, potentially teratogenic drug use, or pregnancy-related disease of his mother. The patient was non-smoker, and physical examination showed longilin, hypoasthenic constitutional type, poor nutritional status (body mass index – BMI, 17 kg/m²), jugular pulse synchronous with carotid pulse, apex shock in the V-th left intercostal space 1 cm outside the midclavicular line, normal blood pressure, at auscultation heart sounds I and II were duplicated, there was a mesosystolic click followed by a short systolic murmur, telesystolic, at the apex, with highest intensity in the V-th left intercostal space, parasternally, and at the xiphoid appendix, accentuated by post-inspiratory apnea. The antero posterior chest radiograph revealed a slight bulging of the right lower and left middle arches with a cardiothoracic index (CTI) at limit, of about 0.44. Electrocardiography showed a sinus rhythm (SR) with heart rate of 60 beats/min, QRS axis at +30⁰, PQ interval = 0.14 s, and prolonged QRS duration (0.14 s in standard leads and 0.18 s in precordial leads) suggestive of right bundle branch block (RBBB) (fig. 1).

Cardiovascular assessment continued with the study of standard sections used for transthoracic echocardiography, 2D mode, color Doppler, continuous and pulsed: parasternal long axis - normal-sized left heart (left ventricle – LV, 39/26 mm, interventricular septum – IVS, 9 mm, left ventricular posterior wall – LVPW, 9.5 mm), while the right heart has dilated cavities (right atrium – RA, 69/39 mm, right ventricle – RV, 79/49 mm), paradoxical IVS movement, the position of the tricuspid valve is diverted to the anterior part and is visible concomitantly with the mitral valve and its excursions are large and closure delayed; anterior mitral valve presents a 6mm telesystolic prolapse and minor mitral regurgitation; apical four-chamber view: septal insertion of tricuspid valve at 26.8 mm inferoapical to the mitral valve, Doppler grade III tricuspid regurgitation (fig. 2); the calculated atrialized RV portion represented approximately 25% of total RV area.

Contrast echocardiography excluded the presence of interatrial communication. Infectious endocarditis prophylaxis, limited physical activity, and low dose diuretics were recommended.

Fig. 1. Case 1 electrocardiogram. SR 60/min, PQ=0,14 s, RBBB

Fig. 2. Case 1 ecocardiography. Apical four-chamber view
**Case 2.** A 34-year-old female patient presenting since childhood repeated fainting spells, intermittent headache, and decreased exercise tolerance was diagnosed at age 14 with Ebstein's anomaly, severe form. The patient was recommended antiplatelet therapy and limited physical exertion; her condition had improved and, later on she carried two pregnancies to term, delivery by cesarean section. This year, the patient's condition worsened again, with rest and exertional dyspnea, chest pain, cyanosis, dizziness, headache and more episodes of loss of consciousness. Emergency craniocerebral computed tomography revealed a moderate diffuse cerebral atrophy. Cardiovascular assessment showed: grade I obesity (BMI 31.5 kg/m$^2$), increased cross-sectional cardiac dullness, sound I diminished, sound II duplicated, systolic murmur grade IV/VI in the III-rd and IV-th left intercostal space; electrocardiogram: 60/min SR, QRS axis at $+30^\circ$, first-degree atrioventricular block (AVB) (PQ = 244 ms), major right bundle branch block, RA and RV overload (fig. 3); echocardiography: low insertion of the tricuspid valve (at 47 mm from the insertion of mitral valv, cusp inserted into the free wall of the right ventricle, separately from tricuspid ring) causing atrialization of a large RV portion (38% of RV area) (fig. 4); left cavities were compressed by the right ones, paradoxical IVS motion; Doppler grade III tricuspid insufficiency and Doppler grade I aortic insufficiency; in the interatrial septum (IAS) foramen ovale was open and had a small bidirectional shunt at color Doppler.

Given the worsening of cardiac dyspnea, presence of syncope, cyanosis and echocardiographic appearance, the patient met the indication for tricuspid valve prosthetic replacement or valvular reconstruction, but until then the continuation of antithrombotic and diuretic therapy was recommended.

**DISCUSSION**

In Ebstein’s anomaly, tricuspid valvular disease is highly variable and causes a wide anatomic, clinical and laboratory spectrum, although the malformation is quite uncommon. The presented cases illustrate this variability. The first patient is a man in which the disease has progressed discretely and passed almost unnoticed until the age of 51 years, the second case is a relatively
young female who, despite two childbirths, has been symptomatic since childhood and last year symptoms worsened so much that surgery was indicated. This prognostic difference in is related to the echocardiographic changes: the apical displacement of the tricuspid valve was more significant in the second case and determined an over 30 percent RV atrialization, considered a sign of severity.

For practical reasons, the studies and anatomical models have gradually been correlated with the echocardiographic findings and classifications of the disease occurred. The best known were the 1988 Carpentier’s anatomo-functional classification (7) and the echocardiographic classification developed by Celermajer in 1994 (8), its goal being to determine, based on the calculation of echocardiographic score, the severity of this condition since birth. According to Carpentier classification, the anomaly may fall into one of four types:

- Type A – the atrialized RV portion is small, anterior leaflet of tricuspid valve is mobile and the septal one is minimally displaced;
- Type B - RV retains functionality, anterior leaflet of tricuspid valve is still mobile, without tethering, but the septal and posterior leaflets are displaced apically more than 25 mm;
- Type C – agenesis of the tricuspid valve posterior leaflet, hypoplasia and severe displacement of the of tricuspid valve septal leaflet, and tricuspid valve anterior leflet partially attached to RV wall, can cause obstruction of the right ventricular outflow tract;
- Type D – the right ventricle is almost completely atrialized, tricuspid valve anterior leaflet is attached to the ventricular wall, forming a second endocardium (5, 7).

Celermajer proposed calculating an echocardiographic ratio of the area of right atrium and the atrialized RV portion compared with that functional RV and left heart, resulting the following classification: grade 1 ratio < 0.5; grade 2 - ratio of 0.5 to 0.99, grade 3 - ratio 1 to 1.49, grade 4 - ratio above 1.5 (4, 8). With reference to these classifications, both presented cases fit Carpentier’ type B, but the calculation of echocardiographic ratio made possible the first patient to be classified as Celermajer grade 2, and the second Celermajer grade 3, indicating a more severe prognosis. The second case also presented a patent foramen ovale. In more than half of the patients with Ebstein’s anomaly an interatrial communication of secundum atrial septal defect type or patent foramen ovale is associated (4).

Although electrocardiography is a classical method that did not seem first line for stratifying the severity of Ebstein’s anomaly, some very recent research support a close relationship between the malformative complex morphology assessed by echocardiography or nuclear magnetic resonance and more electrocardiographic parameters. Assenza GE et al. evaluated several parameters that can be measured on surface ECG: P wave amplitude in DII, PR interval, QRS duration and morphology and correlated them with morphological and magnetic resonance data. For the first time they propose the systematic measurement of S-wave upstroke and revealing a particular pathological aspect present in the terminal portion of the QRS complex or initial ST segment as a small negative wave, below 0.5 mV, aspect called "QRS fractionation". Increased QRS duration
especially in its terminal portion and its fractionation are parameters that correlate well with disease severity, decreased exercise capacity, systolic dysfunction, and RV atrialization (3). In our study, the first patient presented an early repolarization syndrome (known to accompany Ebstein’s anomaly in a variable percentage, between 0 and 44%) (11) and QRS prolongation with RBBB aspect, while the second patient presented AVB grade I and QRS fractionation, elements of severity and poor prognosis (3, 4). For patients with Ebstein disease and asymptomatic preexcitation syndrome, Pappone et al. have proposed the assessment of arrhythmic risk by electrophysiological studies and ablation procedures for high-risk patients (9, 10).

The presented cases had isolated Ebstein’s anomaly, but in the literature numerous complex cases associating other cardiac malformations (pulmonary valve stenosis or atresia, and ventricular septal defects, transposition of great arteries, non-compacted LV) are reported. More cases of Ebstein’s anomaly associated with LV non-compaction are the result of mutations in sarcomere encoding genes, especially MYH7 gene encoding β-myosin heavy chain (11).

Given the discussed severity criteria, the indication for surgery is limited only to the second case. The literature reported medium and long-term favorable results following both prosthetic intervention and plasty consisting in creating a unicusp valve from the anterior leaflet, plicature of the free wall of atrialized portion and posterior tricuspid annuloplasty (12-14).

CONCLUSIONS

Ebstein’s anomaly is one of the rare congenital heart malformations with a highly variable evolutionary spectrum, ranging from fatal cases since childhood to adult asymptomatic cases discovered fortuitously. Patients should be assessed periodically by cardiologists experienced in the diagnosis of congenital heart diseases using electrocardiography, echocardiography, electrophysiology studies and magnetic resonance imaging.

REFERENCES

Ebstein’s anomaly


FACTORs RESPONSIBLE FOR RELATIVE POSITION OF INTERPROXIMAL PAPILLA IN HEALTHY SUBJECTS

A group of Korean researchers conducted a study that examined the factors that can be associated with the appearance of the interproximal papilla. Were examined one hundred and forty-seven healthy interproximal papillae between the maxillary central incisors (male, 74; female, 73; average age, 25.36±7.58 years). A digital photograph and periapical radiograph of the interdental embrasure were taken for each subject using a 1-mm grid metal piece. The recorded parameters were: the amount of recession of the interproximal papilla, contact point-bone crest distance, contact point-cemento-enamel junction distance, cemento-enamel junction-bone crest distance, inter-radicular distance, tooth shape, embrasure space size, interproximal contact area, gingival biotype, papilla height, and papilla tip form. The amount of recession of the interproximal papilla was associated with: 1) increase in contact point-bone crest, contact point-cemento-enamel junction, and cemento-enamel junction-bone crest distance; 2) increase in the inter-radicular distance; 3) triangular tooth shape; 4) decrease in the interproximal contact area length; 5) increase in the embrasure space size; 6) flat papilla tip form. The amount of gingival recession was not associated with the gingival biotype or papilla height. In the triangular tooth shape, the contact point-bone crest distance and inter-radicular distance were longer, the interproximal contact area length was shorter, and the embrasure space size was larger. The papilla tip form became flatter with increasing inter-radicular distance and cemento-enamel junction-bone crest distance. The relative position of the interproximal papilla in healthy subjects was associated with the multiple factors and each factor was related to the others. A triangular tooth shape carries a higher risk of recession of the interproximal papilla because the proximal contact point is positioned more incisally and the bone crest is positioned more apically. This results in an increase in recession of the interproximal papilla and flat papilla tip form. (Kim JH, Cho YJ, Lee JY, Kim SJ, Choi JI. An analysis on the factors responsible for relative position of interproximal papilla in healthy subjects. J Periodontal Implant Sci, 2013; 43 (4): 160-167)

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