CLINICAL AND PARACLINICAL ASPECTS OF EBSTEIN’S ANOMALY - SEVERE FORM IN NEWBORNS

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CLINICAL AND PARACLINICAL ASPECTS OF EBSTEIN’S ANOMALY – SEVERE FORM IN NEWBORNS (Abstract): Ebstein’s anomaly is a rare heart malformation, with a broad spectrum of anatomic abnormalities, leading to different clinical, electrocardiographic, echocardiographic changes and a reserved prognosis. We have described a case of an infant diagnosed with Ebstein’s anomaly – a severe form. Keywords: EBSTEIN’S ANOMALY – SEVERE FORM, PAROXYSMAL SUPRAVENTRICULAR TACHYCARDIA

Ebstein’s anomaly is a congenital malformation of the tricuspid valve in which septal and posterior valves are not attached to the tricuspid annulus, having a spiroid line, shifted to right ventricle apex; and the anterior foil is usually malformed, abnormally attached or adherent to right ventricle. The prevalence of disease is of 5.2 to: 100,000 newborns, representing 0.5% of MCC (1, 2, 3).

The control studies that have been conducted so far suggest the involvement of anomaly more frequently in twins, in children with a family history of MCC and whose mothers were treated with benzodiazepines (6).

80% of patients with Ebstein's anomaly can also develop interatrial communication (7, 8) through which right-to-left shunting of blood that can occur are right-left blood, and 20%: bicuspid aortic coarctation of the aorta, aortic valve atresia, atresia or pulmonary hypoplasia, mitral valve prolapse. The severity of hemodynamic disorders in patients with Ebstein's anomaly depends on the degree of movement and functional status of the tricuspid valve foils. In patients with minimal apical displacement of tricuspid foils, valvular function is normal; while patients with severe displacement foils or abnormal tricuspid valve attachment have an increased pressure in AD and right-to-left interatrial shunting (5).

We will discuss the clinical, electrocardiographic, echocardiographic features and therapeutic management of a newborn diagnosed with Ebstein’s anomaly.

CASE REPORT

We will present the case of a male infant aged 11 days, which is hospitalized by clinical transfer with generalized cyanosis, polypnea, tachycardia and the investigation revealed a systolic murmur detected in maternity.

Family history: the infant is the third
child of an apparently healthy couple; mother aged 31 yrs. and father aged 40 yrs. There are no cases of congenital anomalies or genetic disorders in the family.

**Personal physiological history:** Pregnancy was assessed 5 times by the family physician; mother was admitted to hospital for premature impending birth at 32 weeks. Birth was full term, natural assisted by medical staff in the ambulance, birth weight: 3,400 g, neonatal hypothermia (35.3°C)

**Pathological personal history:** Diagnosis of neonatal hypoglycemia, maternofetal infection and congenital heart malformations is made in the neonatology department.

**Clinical history at admission:** General poor state, cyanotic elastic skin, cold extremities, FA-3/4 cm normotensive, normal chest, symmetrical rib excursion, tachypnea 45/min, SaO₂(-):70%, SaO₂(+):98%, rough MV, without over-added rales, chest area normally conformed, tachycardic rhythmic heart sounds, AV=160/min, systolic murmur degree III / 6 in II intercostal left space. BP right upper limb: 67/47 mmHg, BP right leg 65/41 mm Hg, liver at 2 cm below costal margin, abdomen depression, bowel movements and physiological urination.

Cardio-thoracic radiography shows a global cardiomegaly ICT=0.7, normal pulmonary transparency and free cistodiaphragmatic sinuses (fig.1).

The electrocardiogram shows sinus rhythm of 140/min, electrical axis to the right (+120 degrees), right atrial overload (where P is high, sharp in DII, DIII), major block of right bundle branch (prolongation of QRS=180 msec in pericordial leads) with secondary repolarization changes, BAV degree I (PR interval elongated PR=210 msec) (fig.2).

Fig. 1. Cardio-thoracic radiography global cardiomegaly

![Cardio-thoracic radiography global cardiomegaly](image1)

Fig. 2. Electrocardiogram RS 140/min BRD, BAV

![Electrocardiogram RS 140/min BRD, BAV](image2)

Transthoracic echocardiography was the chosen method for the diagnosis of Ebstein’s anomaly; there are right cavities with increased volume, SIV paradoxal movements. Apical 4C, there appears the apical displacement of the tricuspid septal insertion foil 5.3/5.5 mm/m² body surface area, VD degree of atrialization is over 80%, with compression of the left ventricle, intrarial septal discontinuity, type of oval patent foramen are bidirectional, predominantly right side. Heart failure FE: 38% (fig.3). All elements support Ebstein’s anomaly diagnosis.

Genetic examination revealed a small skull (PC:-1.5 DS), FA large 4/4 cm, nor-
motensive, mild facial dysmorphia.

The infant received spironolactone during hospitalization, patient’s state remaining stationary under treatment.

Fig. 3. Patient S.A.G.: echocardiography (four chambers view) Ebstein’s anomaly severe form

The patient was transferred to the Institute of Cardiovascular Diseases in Targu Mures for possible univentricular corrections.

Tachycardia with wide QRS (possible antidromic lead) appeared during hospitalization, with hemodynamic decompensation; after a parenteral load with Amiodarone, electrical cardio version is attempted but the patient relapses in tachycardia, for this reason a beta blocker was added to the initial treatment, being kept short periods of sinus rhythm. The patient is transferred to Department of Pediatric Cardiology, where he continues treatment and held for surgery.

When was readmitted to Pediatric Cardiology Department, patient’s state was relatively good, dysmorphic facies, elastic skin, generalized cyanosis, moderate hypotonia, slightly tightened vesicular murmur, intercostal and subcostal circulation. Stetacustic heart - rhythmic heart sounds, noise II duplicated, III noise present, systolic murmur degree III/6 parasternal left. Other signs: abdomen depression, liver 2-3 cm below the costal margin, no signs of meningeal irritation.

Electrocardiogram displayed a sinus rhythm of 120 bpm, axis QRS +60 degree, PQ=0.16 sec, right ventricular overload, major block of right bundle branch, without signs of terminal phase QT: 30 sec.

**DISCUSSION**

In Ebstein’s anomaly, variability of tricuspid valve displacement causes diverse clinical, electrocardiographic, echocardiographic changes. Depending on the anatomical changes, Carpentier made this anatomical and functional classification:

- type A: minimum displacement of the septal tricuspid valve, wide, mobile anterior tricuspid valve, small atralized portion of VD, good functional VD;
- type B: displacement of the septal valve of tricuspid and posterior valve by 25 mm, wide, mobile anterior tricuspid valve, without tracking; relatively good functional VD;
- type C: anterior tricuspid valve partially attached to the VD wall, posterior tricuspid valve agenesis, severe hypoplasia of septal tricuspid valve;
- type D: all right ventricle is covered by anterior tricuspid valve, with minimum systolic opening (4,12).

Heart failure is a major factor in the subsequent management of the case. The
Clinical and paraclinical aspects of Ebstein’s anomaly - severe form in newborns

degree of heart failure presented by the infant is supported by the small size of VD, a significant size over 80% of VD atrialization, septal tricuspid valve hypoplasia, left ventricular dysfunction secondary to interventricular septum bulge to the left in diastole, hypoplasia of left heart HTAP (9).

Symptoms were present even at birth with generalized cyanosis, SaO2 (-): 85%, tachypnea and tachycardia; it progressively worsened due to hypoglycemia, neonatal hypothermia and maternal fetal infection.

Although electrocardiography is a classical method which is considered to be the first choice for Ebstein disease severity detection, recent studies support a close relationship between morphological changes of assessed complexes and echocardiographic modifications or nuclear magnetic resonance (10).

Cardiac arrhythmias: Paroxysmal supraventricular tachycardia, ventricular tachyarrhythmias secondary to VD and VS damage are a negative prognostic factor and ablation is indicated using radiofrequency.

In patients with severe tricuspid regurgitation, class III-IV NYHA heart failure, cyanosis, dilation or dysfunction of VD, monocuspe tricuspid valve reconstruction from anterior foil of anterior tricuspid simultaneously with DSA closure are indicated (11, 12, 13).

CONCLUSIONS

Ebstein’s anomaly is one of the rarest congenital heart diseases with highly variable evolutionary spectrum.

Newborns having pronounced cyanosis, heart failure phenomena, cardiac arrhythmias, cardiomegaly with increased cardiothoracic index and severe hypoplasia of the septal tricuspid valve lead to a negative outcome in case of Ebstein’s disease.

REFERENCES

A SYSTEMATIC REVIEW AND META-ANALYSIS OF 18F-LABELED AMYLOID IMAGING IN ALZHEIMER’S DISEASE

Timely diagnosis of dementia removes prolonged uncertainty, unnecessary investigations, delays in initiation of symptomatic treatments, and recruitment of poorly characterized patients into research trials. Definitive diagnosis may only be achieved by histopathological examination of invasive brain biopsy or postmortem tissue, or molecular genetic testing in a minority with inherited dementia. Clinical diagnostic criteria in recent use for Alzheimer’s disease (AD) often fail to differentiate accurately between AD and non-AD pathology with up to 40% of patients diagnosed with non-AD dementias identified as having pathology consistent with AD at postmortem in some series. When considering the pathophysiology of AD, it has been proposed that the presymptomatic phase is characterized by an early rise in amyloid accumulation, followed later by synaptic dysfunction, tau-mediated neuronal injury, reduction in brain volume, and finally emergence of cognitive symptoms, followed by a clinical syndrome of frank dementia. This suggests a sensitive and specific biomarker of brain amyloid deposition, such as amyloid imaging, would be a useful diagnostic tool, perhaps as an adjunct to investigation of cerebrospinal fluid (CSF) biomarkers (including amyloid and tau), cerebral hypometabolism ascertained by 18F-fluorodeoxyglucose positron emission tomography, hippocampal volume, tractography, and clinical cognitive assessments. The most widely studied radiolabeled amyloid ligand, 11C-labeled Pittsburgh compound B (11C-PiB), demonstrates high affinity and selective amyloid binding. But, disadvantages including expensive technique, a radioactive decay half-life of only 20 minutes and a scanning time of 30 minutes, limited its utility in clinical settings. Nowadays, amyloid imaging using three novel fluorine 18–labeled (18F) tracers, 18F-florbetapir, 18F-florbetaben, and 18F-flutemetamol, with a longer half-life than 11C-PiB of 110 minutes has recently been investigated in a number of studies, including phase 2 clinical trials. These agents have recently gained approval for use in clinical practice. The benefits of identifying amyloid in vivo extend to monitoring disease progression, and, with the advent of treatments targeting amyloid deposition, potentially also as a surrogate biomarker of treatment efficacy. In conclusion, this systematic review and meta-analysis has demonstrated favorable sensitivity and specificity of amyloid imaging with novel fluorine tracers in diagnosis of AD, supporting their use as an adjunct in clinical practice (Yeo JM, Waddell B, Khan Z, et al. A systematic review and meta-analysis of 18F-labeled amyloid imaging in Alzheimer’s disease. Alzheimer’s & Dementia: Diagnosis, Assessment & Disease Monitoring 2015; 1: 5–13).

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