Ebstein's anomaly: a single center survey of a rare congenital heart disease

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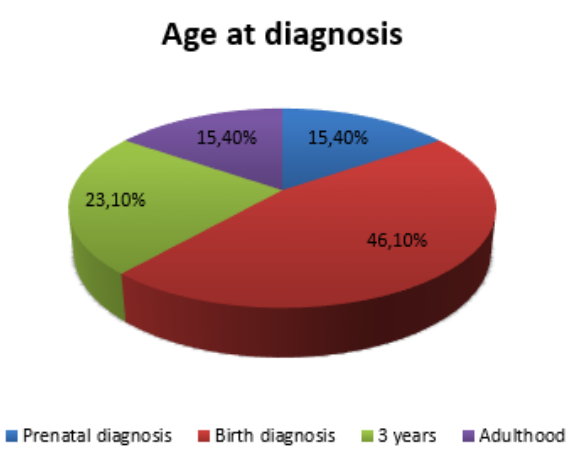
Ebstein's anomaly (EA) is a rare congenital heart defect, which occurs in about 1/200,000 live births. It's currently estimated to account for less than 1% of all cases of congenital heart disease.
Although the pathogenetic mechanism is not yet fully known, in EA there is a lack of delamination of the tricuspid valve (TV) leaflets and subvalvular apparatus. The morphology of the TV in EA is highly variable and this has a major impact on the clinical presentation as well as the outcome.
The aim of this study is to evaluate the clinical and surgical outcome of patients with EA referred to the Pediatric Cardiology Department of the Children's Hospital of Parma from 1975 to the present.

OUR DATA: Thirteen patients were diagnosed with Ebstein's anomaly, 4 (31%) were male, with a mean age at diagnosis of 4 years and 1 month (range 0 - 26 years), **Graph 1**.

The average follow-up period is 14 years and 9 months (range 11 months - 40 years).
In 77% of the patients at least one associated congenital heart disease was described, which, also in our case series, is most frequently represented by an atrial septal defect, Table 1.



Graph 1: age at diagnosis

Associated CHD	*Number of patients*	*Frequency*
Atrial septal defect	8	61,53%
Mitral valve prolapse	2	15,38%
Patent ductus arteriosus	2	15,38%
Ventricular septal defect	1	7,69%
Pulmonary stenosis	1	7,69%

Table 1: associated Congenital heart defect (CHD)

Five patients (38.46%) underwent surgery, at an average age of approximately 7 years (range 4 days -24 years). Indications for surgery were: presence of heart failure, cyanosis and acidosis associated with tricuspid insufficiency, reduced right ventricular function and severe cardiomegaly. Details of the operations are summarised in Table 2.

Type of intervention	*Number of patients*
Valve plane closure and modified BT shunt	1
Glenn operation and TV plastic	1
TV repair according to Carpentier	2
Cone repair	1

Table 2: type of intervention

Cone and Carpentier repair are associated with a significant echocardiographic improvement, in fact the tricuspid insufficiency remained mild to moderate. Another patient is currently waiting for surgery.

Arrhythmias represent a well-know major problem in EA: electrocardiographic signs of pre-excitation were present in three patients. Two of underwent transcatheter ablation of the accessory pathways, in both of them with little success. They currently show partial benefit with anti-arrhythmic therapy (flecainide and propranolol). A third patient is currently awaiting electrophysiological study for possible ablation.
Pregnancy in patients with EA is always a challenge; in our case series, one patient successfully carried a twin pregnancy despite the echocardiographic picture showing severe tricuspid insufficiency and marked enlargement of the right heart sections.