**Case Report**

**Familial Ebstein's Anomaly**

Sreekanth R Shenoy¹, Dipankar Mondal¹, Manasarajan Upadhayay², Jatadhari Mahar³, Leena Das⁴, Saroj Kumar Satpathy⁵

¹Junior Resident, ²Assistant Professor, ³Senior Resident, ⁴Associate Professor, ⁵Professor and Head.
Department of Pediatrics, Sardar Vallabhbhai Patel Post Graduate Institute of Pediatrics (SVPPGIP) and Shri Ramachandra Bhanj Medical College and Hospital (SCBMCH), Cuttack, Odisha-753007, India.

Corresponding Author: Sreekanth R Shenoy

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**ABSTRACT**

Ebstein's anomaly is a congenital cyanotic heart disease with incidence of 0.12 to 0.06 in thousand live births. The primary pathology being down ward displacement of tricuspid valve leaflets. This is a case report of two siblings having ebstien’s anomaly.

**Key Words:** Ebstein's anomaly

**INTRODUCTION**

Ebstein's anomaly is a rare congenital heart disease caused by down ward displacement of tricuspid valve leaflets. This anomaly was first described by Wilhelm Ebstein in 1866 in a report titled, "Concerning a very rare case of insufficiency of tricuspid valve caused by a congenital malformation". [1,2] Most of the cases are sporadic with few cases reports having a familial history suggesting some genetic basis of inheritance. This is a rare case of siblings affected by Ebstein's anomaly.

**CASE REPORT**

A day 2 male child born out of non consanguineous marriage was admitted with respiratory distress soon after birth. He was a term, appropriate for gestational age baby born by LSCS from a 27 year old G2 P2 L0A0 mother. Antenatally there was no history feto toxic medication intake or exanthematous fever. Baby cried immediately after birth and passed urine and meconium on day one of life. There was no history of any repeated vomiting, frothing from mouth, bluish discoloration.

At admission child was sick with severe respiratory difficulty, RR 70/ minute with sub costal retractions with bilateral equal air entry and no adventitious sounds. Heart rate was 140/minute all peripheral pulses were well felt in all four limbs equally. Apex beat was felt on the 4th left inter costal space in mid clavicular line. Loud S1 in tricuspid area, normal S2 with no S3 no S4. There was grade II systolic murmur in tricuspid area. Head to foot examination normal. There was no pallor cyanosis or edema. Abdominal examination revealed soft liver 1.5 cm below the right costal margin. Child developed icterus on day 3 of life.

**Investigations**

Hb-16.3mg/dl total leukocyte count 7500/cc,N68, L26, M6 hematocrit 52% CXR [fig1] showing cardiomegaly and box shaped heart. ECG was showing large p waves. ECHO was suggestive of Ebstein’s anomaly.
The child was stabilized and sent to higher centre for further management.

The first child of the parents was a male child which developed cyanosis on day 1 of life with chest x-ray (figure 2) showing cardiomegaly with boot shaped heart and ECG (figure 3) showing tall p waves and was also diagnosed to have Ebstein’s anomaly by Echocardiography. Baby succumbed on day 7 of life.

DISCUSSION

Ebstein’s anomaly is a rare congenital cyanotic heart disease occurring due to downward displacement of tricuspid valve leaflet. It is a rare disorder with incidence of 1 in 200 000 live birth contributing to <1% of all cases of congenital heart disease. There are heterogeneous genetic factors in Ebstein’s anomaly. Case control studies suggest genetic reproductive and environmental risk factors (e.g. the anomaly is more common in twins, in those with a family history of congenital heart disease, and those with maternal exposure to benzodiazepines). Maternal Lithium therapy can rarely lead to Ebstein’s anomaly in offspring. Most cases are sporadic; familial Ebstein’s anomaly is rare. In a genetic study of 26 families with Ebstein’s anomaly, 93 of 120 first-degree relatives were evaluated. No case of the anomaly was found, but 2 first-degree relatives had ventricular septal defects, and another, who died at 7 months, was said to have had congenital heart disease. Rare cases of cardiac transcription factor NKX2.5 mutations, 10p13-p14 deletion, and 1p34.3-p36.11 deletion have been described in the anomaly. There are six cases of familial Ebstein’s anomaly reported, three of these were siblings, one was a father and daughter, and another was father and son and other one with uncle and nephew. The familial cases of Ebstein’s anomaly suggest that there could be a genetic basis for the disease for which the mode of inheritance is not known. In our case two siblings are affected and there is no history of consanguinity. And no other
family members were affected and possibility of an autosomal recessive transmission could not be ruled out.

REFERENCES


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