

## Case Report

# Intrauterine Detection of Ebstein's Anomaly and Down's Syndrome. Prenatal Diagnosis of a Rare Combination

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*Ebstein's anomaly, although the most common malformation of the tricuspid valve, is a rare disease. Its association with other syndromes and extracardiac anomalies is very rare and has been reported in only a few cases. A case of prenatal diagnosis of Ebstein's anomaly in a patient with Down's syndrome is reported.*

Ebstein's anomaly was described for the first time as an autopsy finding by Wilhelm Ebstein<sup>1</sup> in 1866. The cardiac anomaly is defined as a variable adherence of the posterior and septal leaflets of a redundant tricuspid valve to the interior of the right ventricular wall, with a more caudal implantation of the valve towards the apex. Ebstein's anomaly, although rare, is the most common congenital disease of the tricuspid valve, accounting for 0.5% of the congenital heart diseases<sup>2</sup>. No association with any particular syndrome exists, and extracardiac defects have been rarely reported<sup>3</sup>. Watson, in a study<sup>4</sup> with approximately 500 cases, reported the association of Ebstein's anomaly and congenital cardiac malformations in 48% of the patients catheterized and in 81% of autopsies. Most associated defects were related to the interatrial septum, corrected transposition of the great vessels, and hypoplasia of the right ventricular outflow tract. In the last 10 years, only 3 cases of the association of Ebstein's anomaly and Down's syndrome have been reported<sup>5-7</sup>. We report a case of prenatally diagnosed Ebstein's anomaly associated with Down's syndrome.

## Case Report

The patient is a 39-year-old pregnant female, GII PI, referred at the 35th gestational week to the Unit of Fetal Cardiology of the Instituto de Cardiologia of Rio Grande do Sul/Universitary Institute of Cardiology (UCF-IC/FUC) due to a suspicion of left ventricular hypoplasia on fetal echocardiography, which showed the following: situs solitus; concordant atrioventricular connec-

tion; a significant right atrial enlargement; and dysplastic tricuspid valve, caudally implanted (severe Ebstein's anomaly), with massive systolic regurgitation to the atrialized portion of the right ventricle and to the giant right atrium (fig. 1 and 2). The right ventricle had a reduced size, due to a reduction in its atrialized inlet. The trunk and branches of the pulmonary artery were confluent, had a reduced caliber, with immobile valve and retrograde flow through the ductus arteriosus (functional pulmonary atresia) (fig. 3). The oval foramen was small and had a fixed bulging of the interatrial septum from right to left, and no biphasic flow through it. The left cavities had normal function and morphology, but the interventricular septum was displaced towards the left ventricle. A small pericardial effusion was evidenced. No signs of hemodynamic decompensation were observed. An obstetrical echography revealed a single fetus in the longitudinal position and cephalic presentation, with normal amniotic fluid volume. The compatible gestational age was 35-36 weeks, with an estimated fetal weight of 2610 g in the 50th percentile. No systemic signs of hemodynamic decompensation existed. The patient was followed up until the 38th week, when interruption of the pregnancy was indicated due to signs of fetal congestive heart failure. The newborn was delivered through a cesarean section with APGAR scores of 7 and 7 in the first and fifth minutes, respectively, and weight of 2,680 g. During the first physical examination, Down's syndrome was diagnosed, and later confirmed on a karyotype. The newborn developed cyanosis in the first hours of life, when oxygen therapy was provided to improve arterial oxygen saturation and reduce pulmonary vascular resistance. Despite the support measures, the newborn evolved with progressive aggravation of the cyanosis and metabolic acidosis. On the third day of life, the newborn required mechanical ventilation with FIO<sub>2</sub> of 1.0, and nitric oxide and milrinone were initiated to optimize the pulmonary anterograde flow. The clinical findings progressively improved, evidencing a hemodynamically stable situation. The complications were as follows: jaundice, which required phototherapy from the second day of life onwards, and left atelectasis, which improved with respiratory physiotherapy. The echocardiogram obtained at birth confirmed the diagnosis of Ebstein's anomaly with a dysplastic right atrioventricular valve and downward displacement - below the valvular ring - of the septal and posterior leaflets, causing severe regurgitation. The right atrium was markedly enlarged, and the pulmonary trunk and branches were well developed with anterograde and retrograde flow through a small patent ductus arteriosus. The oval foramen was patent and had a bidirectional flow; the

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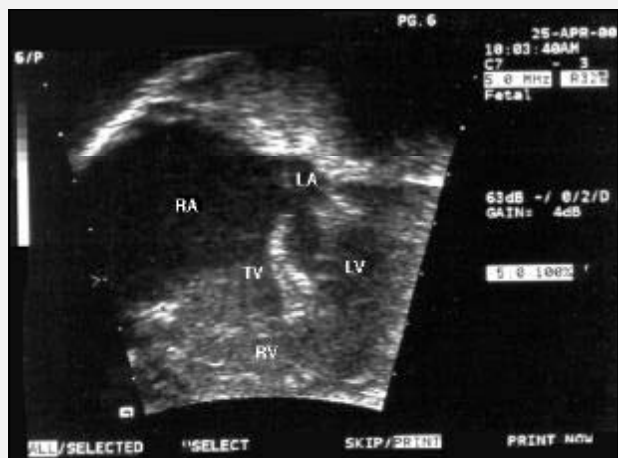


Fig. 1 - Fetal echocardiogram, 4-chamber view, showing right atrial (RA) enlargement with low implantation of the tricuspid valve (TV). LA - left atrium; LV - left ventricle; RV - right ventricle.



Fig. 2 - Fetal echocardiogram with color mapping. Significant tricuspid regurgitation can be observed, with a jet occupying the entire right atrial area. RA - right atrium; LA - left atrium; LV - left ventricle; RV - right ventricle; tric reg - tricuspid regurgitation.

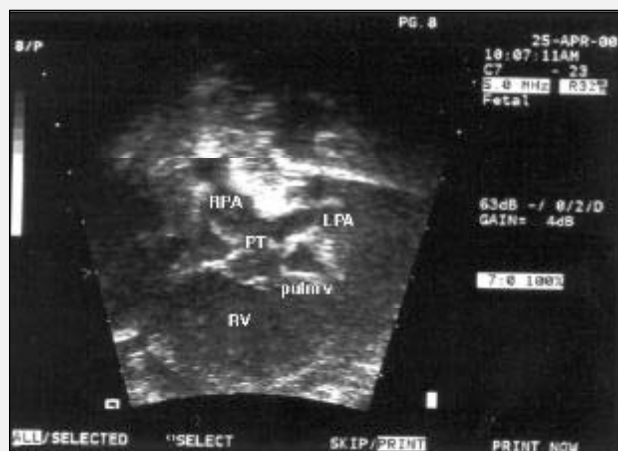


Fig. 3 - Fetal echocardiogram showing that the pulmonary artery connects normally with the right ventricle and that the pulmonary valve is present, although, in the dynamic image, functional pulmonary atresia existed. RV - right ventricle; pulm v - pulmonary valve; PT - pulmonary trunk; RPA - right pulmonary artery; LPA - left pulmonary artery.

area, causing pulmonary hypoplasia. After hemodynamic stabilization on the first days, the patient evolved uneventfully with no new complications and is currently 7 months of age.

## Discussion

Down's syndrome is associated with congenital heart diseases in 50% of cases<sup>8</sup>, the most common associations being septal defects and tetralogy of Fallot. A review of the literature in the past 10 years showed 3 clinical case reports on the association of Ebstein's anomaly and Down's syndrome, and, in only 1, the diagnosis was established in the prenatal period<sup>6</sup>. In the other 2 cases, Ebstein's anomaly was discovered in adulthood, at the age of 55 years<sup>7</sup> and on autopsy<sup>5</sup>. These patients had the mild form of the disease with only a few symptoms.

The case here reported emphasizes the important role played by fetal echocardiography in the prenatal diagnosis of severe heart diseases, such as Ebstein's anomaly, which has an incidence of 1:20,000 in the general population and accounts for 0.5% of the congenital heart diseases found in the neonatal period<sup>3</sup>. Fetal echocardiography may change these figures, because Ebstein's anomaly, if left untreated, is an important cause of fetal death, and, without the establishment of its diagnosis, it cannot be included in the statistics of congenital heart diseases. Prenatal diagnosis and the treatment of these patients, as well as the interruption of pregnancy in the cases with severe and untreatable cardiac failure, may improve the prognosis of these fetuses. The mortality rate of patients with the severe form of the disease in the neonatal period is high, up to 85%, even in large centers, mainly when the symptoms are precocious and surgery is required<sup>9</sup>.

In the present case, the diagnosis of Down's syndrome was not established in the prenatal period, because the mother did not undergo first-trimester nuchal translucency or fetal morphologic ultrasound study in the second trimester, due to her late referral to our institution.

The diagnosis of Ebstein's anomaly in a fetus with Down's syndrome is not frequent, and it is worth reemphasizing the importance of performing the genetic assessment of patients with congenital heart disease. Patients with Down's syndrome should systematically undergo complete cardiological assessment with echocardiography, because of the high incidence of heart disease in those patients. It is worth noting that a cardiological examination with auscultation suggestive of an innocent murmur in a syndromic patient does not mean lack of heart disease, because of the existence of some congenital heart defects that mimic an innocent murmur, such as the mild form of Ebstein's anomaly. This poses great difficulty for the differential diagnosis based only on heart auscultation.

The association of Down's syndrome and Ebstein's anomaly is rare, fetal echocardiography being a powerful tool for assessing fetuses with that combination. Its major function is to evaluate fetal hemodynamic stability, allowing follow-up, treatment, and indication of interruption of pregnancy as a neonatal therapy, when necessary. Prenatal diagnosis is also extremely valuable for providing the pediatrician with ideal conditions for managing those neonates from the delivery on.

left atrium and ventricle were small, but had preserved systolic function. The electrocardiogram revealed right atrial and ventricular hypertrophy, with a normal PR interval and no signs of preexcitation (fig. 4). The chest X-ray showed a greatly enlarged cardiac

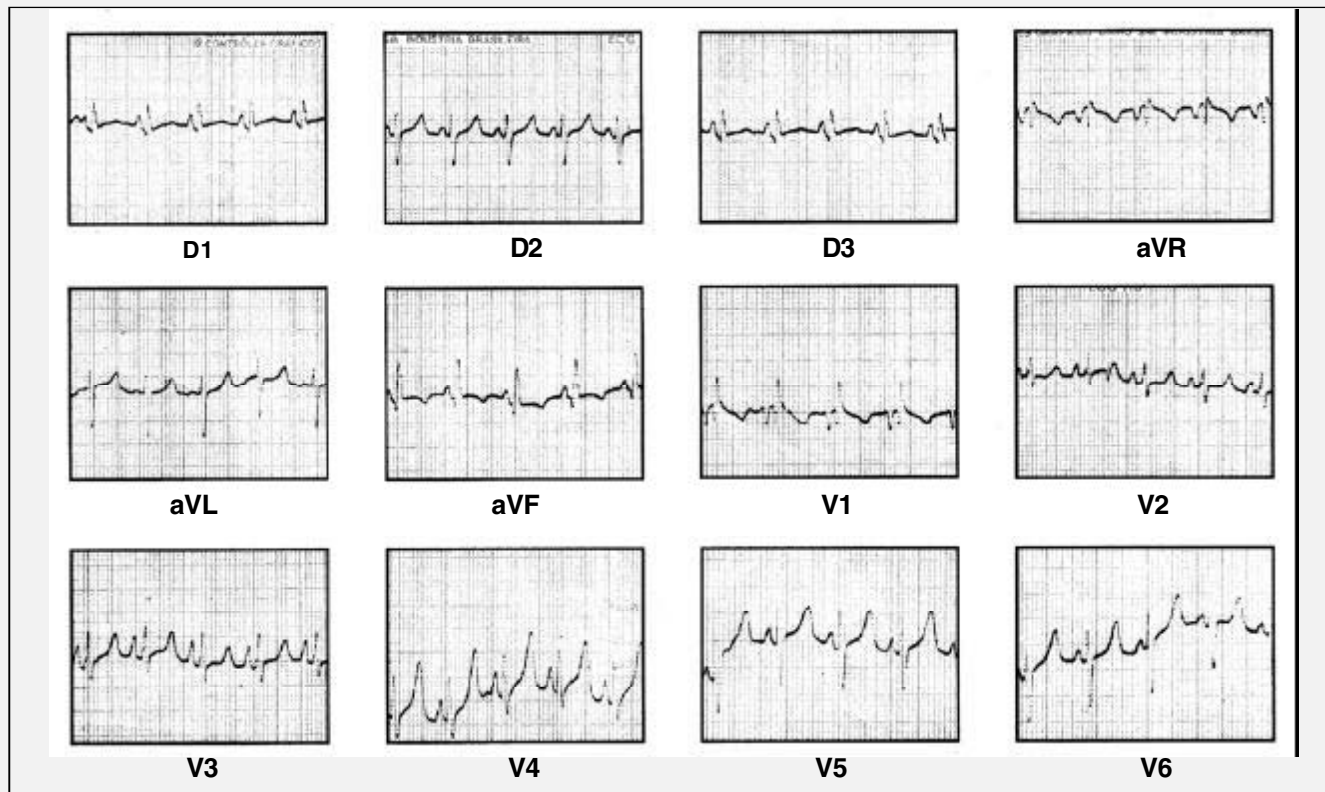


Fig. 4 - Electrocardiogram on the first day of life showing right atrial and ventricular hypertrophy.

### References

1. Ebstein W. Über einen sehr seltenen fall von insuffizienz der valvula tricuspidalis, bedingt durch eine angeborene hochgradige missbildung derselben. *Arch Anat Physiol* 1866; 33: 238-54. *Apud*: Epstein ML. Congenital stenosis and insufficiency of the tricuspid valve. In: Emmanuilides A, Riemenschneider G. Moss and Adams Heart Disease in Infants Children and Adolescents. Maryland: Williams & Wilkins; 1995; 907-21.
2. Epstein ML. Congenital stenosis and insufficiency of the tricuspid valve. In: Emmanuilides A, Riemenschneider G. Moss and Adams. Heart Disease in Infants Children and Adolescents. Maryland: Williams & Wilkins; 1995; 907-21.
3. Davison DL, Bando K, Helfer M et al. Ebstein's anomaly. In: Nichols DG. Critical Heart Disease in Infants and Children. St Louis: Mosby-Year Book, 1995.
4. Watson H. Natural history of Ebstein's anomaly of tricuspid valve in childhood and adolescence. An international co-operative study of 505 cases. *Br Heart J* 1974; 36: 417-27.
5. Johnson CD, Ortiz Colom PM, Sainz de la Peña H et al. Ebstein's anomaly in a patient with Down's syndrome. *Bol Asoc Med P R* 1989; 81: 221-22
6. Silva SR, Bruner JP, Moore CA. Prenatal diagnosis of Down's syndrome in the presence of isolated Ebstein's anomaly. *Fetal Diagn Ther* 1999; 14: 149-51.
7. Venturini E, Musaiio L, Strzzeri R et al. Ebstein's tricuspid anomaly and Down's syndrome. A clinical case report. *Recenti Prog Med* 1992; 83: 556-8.
8. Neill CA, Zuckerberg AL. Syndromes and congenital heart defects. In: Nichols DG. Critical Heart Disease in Infants and Children. St. Louis: Mosby-Year Book, 1995; 987-1012.
9. Mair DD. Ebstein's anomaly: natural history and management. *J Am Coll Cardiol* 1992; 19: 1047-48.