Bilateral Ebstein-like Anomaly with Atrial Septal Defect

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SUMMARY

We report an unusual congenital cardiac anomaly consisting of a downward displacement of all leaflets of both atrio-ventricular valves. In addition to these changes, an atrial septal defect of the secundum type was noted. In light of the extreme rarity of this interesting anomaly complex, we reviewed the literature and compare the morphologic features of the present case with classic Ebstein's anomaly. (*Jpn Heart J* **36**: 119–125, 1995)

Key words: Ebstein anomaly bilateral ASD fetus

EBSTEIN'S anomaly is a cardiac entity/anomaly wherein there is an abnormal origin of one or more leaflets of the tricuspid valve. These do not arise from the annulus fibrosus but instead from the ventricular wall caudal to the atrio-ventricular ring.

Ebstein's anomaly accounts for less than 1% of all heart malformations.¹⁾

In this case report we describe a lesion which, in addition to the usual Ebstein's anomaly, showed an almost mirror image type of change on the left side of the heart. The heart demonstrated situs solitus using the standard morphological criteria for defining the laterality (sideness) of the heart chamber walls.¹⁾ Patent foramen ovale, or atrial septal defect, is associated with classic Ebstein's anomaly in approximately 80 per cent of the cases.¹⁾ In our case, an atrial septal defect of the secundum type was demonstrated at time of postmortem examination.

This is the third case report in the literature that describes a bilateral anomaly and the first of bilateral anomaly and atrial septal defect.

CASE REPORT

This female fetus, delivered prematurely, was the product of a second preg-

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nancy of a 17 year old black mother with no contributory social or medical history, other than a very premature sibling born two years prior.

Family history was positive for chronic hypertension and cardiac surgical procedure, further details of which are not known.

The prior pregnancy and post partum episode ended with the demise within 32 hours of delivery of a very premature neonate weighing 660 g. Permission for autopsy was not granted.

Regarding the present case, the mother was admitted with full dilation of the cervix, with uterine contractions occurring every six minutes. There was no audible heartbeat; the mother could not recall feeling any fetal movement for three days prior to admission. (A previous perinatal ultrasonographic screening done at eighteen weeks of gestation showed normal, appropriate for age, developmental parameters). Shortly after the admission a macerated female fetus was delivered. Permission for autopsy was granted.

Pathologic findings: At necropsy, the fetus weighed 830 gms, measured 33.5 cm crown-heel length. The gestational age was estimated to be 27 weeks by internal organ development and skeletal parameters. Maceration was grade II–III/IV (epidermal peeling and looseness of joints) with accompanying marked visceral autolysis, which precluded chromosomal analysis. No significant external anomalies were identified. The internal organs, apart from their autolytic softness were of normal size and shape. There was situs solitus and the cardiac silhouette (as seen in situ) was relatively unremarkable.

Microscopically the following findings were noted: the lungs revealed an early saccular phase of development with some maturational lag of the vasculature, especially of the medium and small sized arteries. Additionally, there was mild to moderate perivascular sclerosis. The liver showed a few scattered parenchymal psammomatous calcifications of uncertain etiology. There was prominent sub-acute and chronic thymic involution.

Placental findings were as follows: early third trimester changes, acute chorioamnionitis, meconium staining, focal villous sclerosis and intervillous microcalcifications. There was focal obliteration of villous vessels. A focal villitis was noted. The umbilical cord showed three blood vessels on cross-section and no significant funisitis.

Cardiac pathologic findings: The heart weighed 5.3 g (normal weight 6.3 ± 1.8 g, for 750 g body weight). The heart, which was four chambered, was characterized by situs solitus, atrioventricular and ventriculo-arterial concordance. The left and right ventricular walls were 5 mm and 1.5 mm thick, respectively.

There was mild to moderate atrialization of both ventricles: the left atrialized segment measured 3.5 mm of a total ventricular length of 12 mm; the

Figure 1. Gross photograph of the pinned “clamshelled” heart (posterior AV aspect) showing atrialization of both ventricles. The atrialization is best seen on the left and right margin of the heart in this view. (see arrows)

Figure 2. Gross photograph of the heart. This view centers on the atrial septum as seen from the right side. The incomplete membrane of the atrial septal defect of secundum type is clearly evident. (see arrow.) Two smaller membrane defects are not visualized in this photograph. One pin marks the center of the right ventricular atrialized segment. (See larger arrow.)

right atrialized segment measured 2.5 mm of a total ventricular length of 8 mm (Figures 1, 2, 3, 4). The valve circumferences were as follows: tricuspid valve 21 mm, pulmonic valve 8 mm, mitral valve 19.5 mm, aortic valve 7 mm.

Individual valve leaflets of the tricuspid and mitral valves were only mildly dysplastic and arose at the same level caudad to each annulus fibrosus. The aortic and pulmonic valves showed normal architecture.

An atrial septal defect of the secundum type was identified. It was made up of three separate component defects. The largest of these was a lenticular defect

Figure 3. Schematic line drawing of the right sided AV aspect of the heart showing the atrialization of the right ventricle. The solid line shows the actual level of the attachment of the tricuspid valve. Note: All cusps arise at the same level. (See arrow). The dashed line above it shows the normal level of valve attachment. The ASD is demonstrated.

Figure 4. Schematic line drawing of the left sided AV aspect of the heart showing the atrialization of the left ventricle. The solid line shows the actual level of the attachment of the mitral valve. Note: All cusps arise at the same level. (See arrow.) The dashed line above it shows the normal level of valve attachment. The ASD is demonstrated.

of the free edge of the septum primum; the other defects were of pinhead and pinpoint size, respectively, and were located adjacent to the largest one (Figures 3 and 4).

No major inlet or large artery anomalies were seen. There was no appreciable foreshortening of the outflow tracts. Pulmonary venous return showed

unremarkable morphology.

DISCUSSION

This case, to our best knowledge, is the third reported of bilateral Ebstein-like anomaly and the first with a concomitant atrial septal defect. Several excellent reviews of Ebstein's anomaly are available in the literature.^{2,3,4,5)} One previous case of Ebstein's anomaly of the tricuspid and mitral valves has been described by Dusmet et al, but their case did not demonstrate any other pathologic cardiac malformations.⁶⁾ The Dusmet et al report documents important clinical data, including physical/auscultatory findings, electrocardiographic, echocardiographic and Doppler studies.⁶⁾

We compared the morphologic features of our case with those of the case of Dusmet and co-workers and found some significant differences. Our heart (Figures 1, 2, 3, 4) showed uniform caudad displacement of all valve leaflets in relation to each other in both the mitral and tricuspid valves (3.5 mm on the left, 2.5 mm on the right), whereas the cited authors' heart showed downward displacement of the septal and posterior cusps on the right side. On the left side their specimen's mitral valve showed displacement of the posterior valve leaflet to the upper limit of the trabecular portion of the left ventricle. The case of Dusmet et al showed significant dysplasia of the cusps; our case showed only mild dysplastic changes.

The presence of an atrial septal defect makes our case more similar to the classic Ebstein's cases, in that the majority of those show either a patent foramen ovale or an atrial septal defect. The actual numbers vary but the generally agreed upon incidence range is 42–97%.^{2,6)} The Dusmet et al case revealed other dysmorphic features including hypertelorism, retrognathia, widely spaced nipples, stiffness of elbows and knees, left cryptorchidism and slight epispadias. There was hypertrophic stenosis of the pylocalyceal junction and moderate right-sided hydronephrosis. Our case showed none of these features.

Another report documents a case of a bilateral Ebstein-like lesion in association with hypoplasia of the ascending aorta.⁷⁾

The clinical problems of Ebstein's anomaly are related to the individual component lesions making up the complex of the malformations.^{1,2,5,6)} Ebstein's anomaly is most likely the effect of the lack of support of A-V sulcus tissue which is continuous with the tissue of the forming valve leaflet.⁸⁾

The Ebstein-like anomaly of the left side is probably so uncommonly identified because the developing mitral valve could still be supported by the left ventricular myocardium.⁸⁾

Severe dysplasia of the valve leaflets will often be manifested as sail or cone

shaped leaflets, in which the edges are often rolled. Chordae tendineae may be foreshortened or even absent in marked dysplasia.⁴⁾ Part or all of the valve cusp may be plastered from the usual level of origin (at the level of the annulus fibrosus) to the level where the free edge of the cusp leaflet appears to originate. There may be a superimposed mitral prolapse.⁹⁾ Other mitral valve anomalies of the orifice, leaflets, cords and papillary muscles may be seen in association with Ebstein's anomaly.¹⁰⁾ The thin right atrium and atrialized ventricle provide a temporary reservoir for the blood volume which cannot be ejected into the lungs. Moreover, the presence of ASD acts to relieve some of the congestive symptomatology. Atrialization of the ventricle or ventricles affects the function of both the atria and ventricles. Logically, if the degree of atrialization is severe, the mechanical pump function of the ventricular muscle is diminished because of decreased mass and altered configuration. Aneurysm-like areas may be present especially if the atrialized segment is fibrotic or devoid of muscle. Finally, conduction system defects are well recognized in Ebstein's anomaly and have been documented in the Dusmet et al case.⁶⁾

The rare isolated case of left sided Ebstein malformation may be responsible for pulmonary venous obstruction and pulmonary hypertension.^{4,11)} A better understanding of the total complex of bilateral Ebstein-like anomaly with atrial septal defect will come as future reports document the clinical malformation in the living patient with the clinically developed syndrome. On the other hand, autopsy documentation of anomalous fetuses is very important. Special emphasis should be placed on the cardiac development to help pinpoint the triggering event or conditions, and thus, aid in the delineation of the entity's in utero progression, especially its valvular dysplastic component. Another equally important study will be to ascertain the presence or absence of associated anomalies of the cardiovascular and other systems and to recognize possible external dysmorphisms.

The future studies on neonates, infants, and older individuals should include detailed physical, chromosomal/genetic, functional, imaging and electrocardiographic studies as well as appropriate medical and surgical therapeutic techniques.

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