Ebstein's anomaly in persistent common atrioventricular canal

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SUMMARY This report documents 2 patients who presented with cyanosis early in life. In both instances the necropsy showed the simultaneous occurrence of an incomplete type of persistent atrioventricular canal and an Ebstein's malformation of the 'tricuspid' valve component. In the first patient no clinical investigations were done. In the second patient the electrocardiogram was consistent with persistent atrioventricular canal and the angiocardigram showed the characteristic goose-neck deformity. Moreover, an abnormality of the right cardiac contour was seen, which in retrospect was correlated with the right-sided Ebstein's malformation. Ebstein's anomaly may significantly alter the clinical and haemodynamic profile of atrioventricular canal and should be considered in atypical cases. The presence of Ebstein's anomaly will complicate surgical repair of the atrioventricular canal.

Persistent common atrioventricular canal may occur in association with a wide variety of additional congenital heart malformations. However, the simultaneous occurrence of persistent common atrioventricular canal with Ebstein's anomaly of the tricuspid part of the valve is extremely rare. As far as we are aware only 2 cases have been described to date (Kilby *et al.*, 1956; Lev *et al.*, 1961). In both instances an incomplete form of atrioventricular canal was present and the Ebstein's anomaly was recognised during operation and at necropsy, respectively.

It is our purpose to document 2 cases of incomplete atrioventricular canal associated with Ebstein's anomaly of the tricuspid component. In one of these cases an angiographic-pathological correlation was made, a documentation which has not been published to date.

Report of patients

CASE 1

This male patient was born after an uneventful pregnancy and delivery; shortly after birth he developed respiratory distress accompanied by episodes of cyanosis. A heart murmur was noted.

On admission he showed signs of severe heart failure. No further investigations could be performed and the child died at 5 days of age.

Necropsy showed major abnormalities confined to the heart and lungs. The heart was normally positioned in the thorax and had a normal arrangement with respect to segmental connections. Both atria were enlarged. There was an incomplete type of common atrioventricular canal, with a large defect of the primum type (Fig. 1). On the left side the septal leaflet showed a cleft separating an anterior from a posterior component. Both components originated directly from the crest of the septum, not permitting an interventricular communication (Fig. 1B). The valve was otherwise normally inserted into the left-sided annulus fibrosus. The right-sided valve leaflets, on the other hand, showed an additional abnormality consistent with that described in Ebstein's anomaly of the tricuspid valve. The septal leaflet of the right-sided component showed distinct downward displacement, starting at the site where the left-sided cleft reached the ventricular septum. The line of 'origin' of the displaced valve leaflet took a course posteriorly and apically, close to the trabecula septomarginalis, extending on to the inferior wall of the right ventricle, where it gradually veered back towards the atrioventricular annulus reaching it at the site of the margo acutus (Fig. 1A). A large atrialised part of the right ventricle was thus created (Fig. 1A). Moreover, part of the dis-

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Fig. 1 Case 1. (A) Opened right side of the heart. There is obvious distal displacement of the septal and inferior attachments of the tricuspid valve component (dotted line); compare with site of annulus fibrosus (solid line). A large atrialised part of the right ventricle is formed (between dotted and solid lines). A bare area (between arrows) is present between the displaced septal leaflet and the anterior 'tricuspid' component. Note that part of the inferior leaflet is muscularised (asterisk). A large primum type atrial septal defect is present, through which left heart structures are visible. CS, ostium of coronary sinus; FO, fossa ovalis. (B) Opened left side of the heart. The mitral valve component reveals a deep cleft (arrows), reaching to the crest of the ventricular septum and dividing this valve component into an anterior and a posterior part.

placed valve leaflet was muscularised. A 'bare area' was present between the displaced septal leaflet and the anterior 'tricuspid' component. The latter took its 'origin' in a normal fashion from the annulus fibrosus, but the valve leaflet itself was curtain-like with direct insertions into right ventricular muscular trabeculae. The right ventricular infundibulum was normal, whereas the left-sided outflow tract was much narrowed because of the deficient ventricular septum and the firmly attached anterior leaflet. The great arteries showed no abnormalities; the ductus arteriosus was probe patent.

The lungs were congested and microscopical examination showed distension of pulmonary veins with a suggestion of 'arterialisation'.

CASE 2

Since birth this girl had had feeding difficulties caused by cheilognathopalatoschisis. The child was admitted to hospital at the age of 2 months because of progressive dyspnoea and cyanosis. Physical examination showed slight cyanosis at rest, but no signs of heart failure. Auscultation disclosed normal first and second heart sounds; there was no

splitting of the second sound. A high frequency holosystolic murmur, grade 2/6, was noted. Both femoral arteries pulsated well. The plain chest film showed an egg-shaped, slightly enlarged heart with a left-sided aortic arch, a normal pulmonary segment and normal pulmonary vascular markings with no pulmonary venous congestion. The electrocardiogram showed a sinus tachycardia of 150 beats/min and signs of left ventricular hypertrophy. The electrical axis in the frontal plane was directed to the left. At cardiac catheterisation the right ventricle and pulmonary artery were not reached; there was a right-to-left shunt at atrial level with an arterial saturation of 73 per cent. Angiography showed a goose-neck deformity of the left ventricular outflow tract (Fig. 2A) and a small left-to-right shunt at ventricular level (Fig. 2B). Contrast injection into the right atrium showed an enlarged right ventricle, while the lateral projection of this injection revealed an indentation in the inferior border of the right ventricular cavity, dividing it into a smoothly outlined basal part and a trabeculated apical part (Fig. 3A). A normally positioned pulmonary trunk was visualised. At that time the malformation was not considered operable and the child was treated

Fig. 2 Case 2. Left ventricular angiogram. (A) Antero-posterior view revealing the 'goose-neck' deformity (arrows) of the LV outflow tract. (B) In the lateral view a small left-to-right shunt (between arrows) is visible.

medically. However, she gradually developed progressive heart failure and died at the age of 2 years.

Necropsy showed that the major abnormalities were confined to the heart and lungs. There was an incomplete form of persistent atrioventricular canal, with enlargement of both atria. The atrial septum showed a huge defect of the primum type, in addition to a fenestrated fossa ovalis (Fig. 4A and B). The left-sided atrioventricular valve showed an incomplete cleft in the septal leaflet (Fig. 4B). The outflow tract of the left ventricle was narrowed

with a high insertion of the anterior part of the septal mitral valve component, in accordance with the clinical observation of a 'goose-neck' deformity (Fig. 2A and 4C). The right-sided atrioventricular valve showed distinct downward displacement which was present over the septum and extended on to the inferior wall (Fig. 4A). The valve 'origin' came back to the annulus fibrosus at the level of the *margo acutus*. The anterior leaflet was normally inserted but curtain-like with localised muscularisation. A bridge of valve tissue was present between the displaced septal part, where it attached to the crest of the ventricular septum, and the anterior leaflet, thus creating a superior small 'extra' ostium with a diameter of 10×5 mm. Because of the abnormal 'origin' of the septal and inferior parts of the 'tricuspid valve' a large atrialised part of the right ventricle was formed. In retrospect it is clear that the atrialised part of the right ventricle corresponds with the smooth walled segment of the right ventricle seen on the angiogram (Fig. 3). A small muscular ventricular septal defect was present through which the left ventricle was connected to the inflow part of the right ventricle, immediately apical to the displaced septal leaflet (Fig. 2B and 4C). Both right and left coronary arteries had their ostia positioned above the right anterior aortic cusp. The left coronary artery took its origin from the posterior ostium and coursed posteriorly to the aorta after which a normal distribution pattern occurred.

The lungs were congested. Microscopical examination showed that the pulmonary veins had a very

Fig. 3 Case 2. Correlation between right heart angiogram and pathological anatomy. (A) Lateral view of the right-sided angiogram. The inflow part of the right ventricle shows a smooth inferior border (arrows) separated by an indentation from a trabeculated apex (asterisk). (B) The opened right side of the heart of this patient, viewed in similar direction. The smooth inferior border of the inflow part of the right ventricle (arrows) is caused by atrialised right ventricle, from Ebstein's anomaly of the tricuspid valve component. RAA, right atrial appendage.

Fig. 4 Case 2. (A) Opened right heart showing a large atrialised part of the right ventricle caused by distal displacement of the septal and inferior attachments of the tricuspid valve component (dotted line); compare with site of annulus fibrosus (solid line). A superior 'extra' ostium, barely visible (arrow), is created because of a bridge of valve tissue between the septal and anterior 'tricuspid' valve components. There is a large primum type atrial septal defect through which left heart structures are visible. The fossa ovalis is fenestrated. CS, ostium of coronary sinus; ICV, ostium of inferior caval vein. (B) The left side of the heart reveals a shallow cleft (arrows) in the 'mitral valve', a large primum type atrial septal defect and a fenestrated fossa ovalis. A muscular ventricular septal defect is present (VSD; compare with Fig. 2B). PPM, posterior papillary muscle. (C) Outflow segment of the left ventricle with high insertion of the anterior part of the septal mitral valve component (arrows), contributing to a narrowed outflow tract (compare with Fig. 2A). VSD, ventricular septal defect; AoV, aortic valve.

hypertrophic media with 'arterialisation' of the wall. The pulmonary arterioles were slightly thin walled. Fibrosis was present in septa and sub-pleurally.

Discussion

The combination of Ebstein's anomaly of the 'tricuspid' valve with persistent common atrio-ventricular canal is rare. To the best of our knowledge this condition has been reported only twice previously. Kilby and associates (1956), in a clinical study on Ebstein's malformation, reported a 13-year-old girl (their case 9) with this combined malformation. Catheterisation had shown a large atrial septal defect with a predominant left-to-right shunt, while the course of the catheter suggested a low lying defect. Surgical exploration disclosed an incomplete form of persistent atrioventricular canal, in addition to Ebstein's anomaly of the septal 'tricuspid' valve component. The surgeon palpated a narrowed mitral valve, which might have contri-

buted to the presence of a large left-to-right shunt. The child died, but necropsy was not permitted.

The second published report is that by Lev and associates (1961). They described the association of a 'Fallot-type' ventriculoarterial relation with persistent common atrioventricular canal. In one of their patients, a 1-month-old male infant (case 1), an Ebstein-like malformation of the right-sided valve component was found at necropsy. The valve showed displacement of the septal and inferior parts and dysplasia of the anterior leaflet. Again, an incomplete form of persistent atrioventricular canal was present. The child had been admitted to hospital because of dyspnoea and cyanosis since birth.

CLINICAL IMPLICATIONS

The combination of Ebstein's anomaly of the 'tricuspid' valve with persistent common atrioventricular canal is of practical significance. Of the two conditions it is most likely that the persistent atrioventricular canal will be diagnosed first. However, patients with persistent atrioventricular canal nearly

always present with signs of a left-to-right shunt; cyanosis as an early symptom is rare (Ongley *et al.*, 1976) and may indicate an additional right heart problem. The patients described by Lev and associates (1961), presenting with a 'Fallot-type' morphology, are perfect examples in this respect and we suggest that Ebstein's anomaly of the tricuspid valve component should also be included among the possible diagnoses. In atrioventricular canal defects severe symptoms in infancy are always associated with one or more of the following factors: a large left-to-right shunt, severe mitral regurgitation, and significant associated cardiac malformations (Ongley *et al.*, 1976). Therefore, if an infant with an atrioventricular canal defect has severe and progressive symptoms, in the absence of a large left-to-right shunt or severe mitral regurgitation as in our second patient, complicating malformations should be suspected and looked for.

Recognition of these features is of particular significance since when 'persistent common atrioventricular canal' is diagnosed the main interest is focused on the left side of the heart. For diagnostic purposes left ventricular angiograms are mandatory while for surgical correction left ventricular dimensions and detailed visualisation of the 'mitral' valve component are vital. A complete and detailed study of the right side of the heart is considered unnecessary for either purpose. Even with reflux into the right atrium further study of the right-sided valve components is unnecessary, since it has been shown that in the majority of cases the regurgitation

is caused by leakage of the left-sided atrioventricular valve.

Our present observations re-emphasise the fact that Ebstein's anomaly of the right-sided valve may coexist with the incomplete form of persistent atrioventricular canal and that this abnormality may play an important role in altering the haemodynamics and symptoms. As an example both infants were cyanotic early in life, a very uncommon finding with incomplete atrioventricular canal. It is obvious, moreover, that the presence of Ebstein's anomaly will complicate surgical repair of atrioventricular canal.

We are indebted to Dr Niehuisen, Stichting St, Elisabeth Ziekenhuis, Sluiskil, for the clinical data on case 1.

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