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Original Article

Clinical and echocardiographic features of Ebstein's malformation in Sudanese patients

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Abstract Ebstein's malformation is a rare congenital cardiac malformation, accounting for about 0.5% of all congenital cardiac lesions. We report our experience with the anomaly as encountered at the Sudan Heart Centre from July 2004 to April 2005. Diagnosis was based on the echocardiographic demonstration of displacement of the septal leaflet of the tricuspid valve towards the ventricular apex by greater than 8 millimetres per metre squared. *Results:* In a period of 10 months, we identified 12 patients with the malformation, this number constituting 2% of all patients seen with congenital cardiac disease. The age ranged from 2 weeks to 35 years. Of the patients, half were asymptomatic, while the other half presented with congestive cardiac failure, 4 of these having cyanosis in addition to heart failure. Associated diseases included chronic renal failure, impaired hearing, stunted growth, and developmental delay. Electrocardiographic abnormalities included peaked and tall P waves, seen in four-fifths, an Rsr pattern, first degree atrioventricular block, atrial fibrillation, and Wolf–Parkinson–White syndrome, including the Mahaim pattern of pre-excitation. Associated abnormalities diagnosed echocardiographically included atrial septal defects, prolapse of the leaflets of the mitral valve, left ventricular dysfunction, atrial septal aneurysm, pulmonary valvar stenosis, and pericardial effusion. *Conclusions:* Ebstein's malformation was seen four times more frequently at the Sudan Heart Centre when compared to the average frequency reported in the Western literature. Many of the associated diseases encountered in Sudan had not previously been reported.

Keywords: Tricuspid valve; incidence; congenital heart disease

EBSTEIN'S MALFORMATION OF THE TRICUSPID valve is a rare congenital heart disease, with a reported incidence of 1 for every 210,000 live births. It is said to account for about 0.5% of all congenital cardiac diseases.[1,2] Familial incidence, and an association with Down's syndrome, has been reported.[3–5] To our knowledge, there have been no reports about the clinical and echocardiographic features as seen in African patients. We describe our experience with this disease in Sudan, where we observed a strikingly high frequency and unreported associations.

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Patients and methods

For the purposes of our review, we included all patients with Ebstein's malformation seen at a tertiary cardiac center, namely the Sudan Heart Centre, from July 2004 to April 2005. The patients were evaluated by clinical examination, chest X-ray, and electrocardiograms. Complete cross-sectional and Doppler echocardiographic studies were performed for each patient using the MEGAS (Esaote) machine. Cardiac catheterization was undertaken in those patients when there was doubt about the echocardiographic diagnosis. Echocardiographic diagnosis was based on the finding of apical displacement of the septal leaflet of the tricuspid valve by more than 8 millimetres per square metre, with or without tricuspid valvar regurgitation, abnormal attachment of the septal leaflet to the ventricular septum, an elongated and whip-like antero-superior leaflet, and dilation of the right atrium.

Associated anomalies were sought, and reported when present. We excluded from consideration patients with Ebstein's malformation in the context of congenitally corrected transposition.

Results

The characteristics of the patients identified are shown in Table 1. Over a period of 10 months, we identified 12 patients with the echocardiographic criterions justifying the diagnosis for Ebstein's malformation. There were 7 males and 5 females. During this period, we performed 700 echocardiographic studies. Of this load, 595 patients (85%) had congenital cardiac disease, 70 (10%) had rheumatic disease, and the remainder had other cardiac diseases. Ebstein's malformation, therefore, accounted for 2% of all patients with congenital cardiac disease. The frequency of other congenital cardiac malformations in our patients was similar to that described in the literature. The age of the patients with Ebstein's malformation ranged from 2 weeks to 35 years, with means and medians of 10 years. We were unable to elicit any history of maternal ingestion of anti-psychiatric drugs.

Mode of presentation. Half of the patients were asymptomatic, with one being referred for routine echocardiography prior to preparation for renal transplantation. Of the others, 4 were referred because of heart murmurs, and one because of cardiomegaly identified on the chest X-ray. The remaining 6 patients presented with congestive heart failure, 5 being in Grades III or IV of the classification of the New York Heart Association, and 1 in class II. Of these patients, four presented with cyanosis in addition to cardiac failure.

Associated diseases. Chronic renal failure, associated with haematuria and familial deafness, was seen in our first patient. Our eighth patient had impaired hearing associated with delayed milestones. Severe growth retardation was seen in 5 boys. All had weights and heights below the third centile. Jaundice due to hepatitis C was seen in one patient. Mild right-sided hemiparesis developed in our fourth patient during follow-up, while abdominal tuberculosis was diagnosed in our second patient.

Electrocardiogram. Peaked and tall Himalayan P waves were seen in 11 patients (91%). In 3 patients, we found an Rsr pattern, while first degree atrioventricular block was seen in 2, and atrial fibrillation in 1. Wolf-Parkinson-White was found in three patients, 1 with pre-excitation and 2 with delta waves but a normal PR interval, the so-called Mahaim type of pre-excitation.

Echocardiogram. The mean index for displacement of the tricuspid valve was 17.7, this not being statistically different for symptomatic as opposed to asymptomatic patients, with one of the latter patients having an index of 43 (Fig. 1). In 11 patients, the hinge points of both the septal and mural leaflets of the valve were displaced within the inlet component of the right ventricle, while in the tenth patient, the septal leaflet had not delaminated from the muscular ventricular septum. In 2 patients, we observed grossly abnormal distal attachments of the anterosuperior leaflet, and in both the valvar regurgitation was severe. In the remaining patients, regurgitation was moderate to severe in 6, and mild in the others. Tethering of septal leaflet to the septum was seen in 5 patients. A large redundant and whip-like antero-superior leaflet, with abnormal distal attachments, was seen in 6 patients. The right ventricle was considered unduly hypoplastic in 3 patients. Mitral valvar prolapse, with moderate regurgitation and left ventricular dysfunction, was seen in 2 patients. Other associated anomalies included an atrial septal defect at the oval fossa in 4, a fenestrated oval fossa with an aneurysm of the flap valve in 1, and pulmonary valvar stenosis in another. Pericardial effusions were seen in 3 patients, needing aspiration in one.

Cardiac catheterization was carried out in our third patient. The mean right atrial pressure was measured at 15 millimetres of mercury, and mean pulmonary arterial pressure was 20 millimetres of mercury. The atrialized right ventricle was well visualized, albeit with a right atrial pressure tracings. Severe tricuspid regurgitation, and severe dilation of the right atrium, were noted.

Management and follow-up. The third patient underwent replacement of the tricuspid valve in another centre, and was lost to our follow-up. The other symptomatic patients were commenced on anti-failure medications, in the form of diuretics and inhibitors of angiotensin, and have been discussed for future surgical treatment. The patient with atrial fibrillation was treated with beta-blockade. The fourth patient developed mild right-sided hemiparesis that improved over the course of a few days.

Echocardiography did not show thrombus in either the right or left atrium, and she was in sinus rhythm on the electrocardiogram. She was started on warfarin. We have scheduled two patients for transcatheter closure of their atrial septal defect.

Discussion

The frequency of Ebstein's malformation as seen in our centre, accounting for 2% of all patients seen with congenital cardiac disease, is four times the average reported frequency in findings similar to ours. As far as we are aware, there are no previous reports about the frequency in Africa, but an investigation

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Table 1. Showing patients' characteristics.

No	Sex	Age	Presenting symptom	NYHA	Clinical/ association	Cardiothoracic ratio on chest X-ray	Electrocardiogram	Echocardiogram
1	M	10 years	Asymptomatic	I	Chronic renal failure. Impaired hearing. Stunted growth	50%	Right atrial enlargement	DI: 18 Mild tricuspid regurgitation
2	F	15 years	CHF Anasarca	III	Tuberculosis	80%	Right atrial enlargement Rsr	DI: 15 Severe tricuspid regurgitation. Hypoplastic right ventricle. Massive pericardial effusion
3	M	7 years	CHF Cyanosis	III	Stunted growth/ dysmorphic	80%	Right atrial enlargement Rsr First degree block	DI: 8 mm/m^2 Severe tricuspid regurgitation. Small atrial septal defect
4	F	35 years	Asymptomatic	I	Hemiparesis noted on follow-up	50%	Right atrial enlargement. First degree block	DI: 20 Large atrial septal defect. Mild tricuspid regurgitation
5	F	10 years	CHF	IV	Normal growth	80%	Right atrial enlargement	DI: 15 Severe tricuspid regurgitation. Left ventricular dysfunction. Mitral valvar prolapse with moderate regurgitation
6	M	16 years	Asymptomatic	I	Jaundice (Hep C). Stunted growth	50%	Atrial fibrillation	DI: 12 Mild tricuspid regurgitation
7	M	2 weeks	CHF Cyanosis	IV	Cyanosis improved on follow-up	80% Oligemic lungs	Right atrial enlargement. Wolff–Parkinson–White Rsr	DI:10 Severe tricuspid regurgitation
8	M	3 years	Asymptomatic	I	Murmur. Hearing impairment. Stunted growth. Delayed milestones	50%	Right atrial enlargement. Right ventricular hypertrophy	DI: 14 Mild tricuspid regurgitation. Large atrial septal defect. Pulmonary valvar stenosis.
9	M	6 years	Asymptomatic	I	Stunted growth	55%	Left axis deviation. *Mahaim type pre-excitation*	DI: 43 Mild tricuspid regurgitation. Small atrial septal defect
10	F	10 years	Cyanosis and CHF	III	Normal growth	50%	Right atrial enlargement. Mahaim *type pre-excitation*	Septal leaflet completely tethered to septum. Severe tricuspid regurgitation. Small atrial septal defect
11	F	8 years	Asymptomatic	I	Normal growth	50%	Right atrial enlargement Rsr	DI: 35 mm Fenestrated atrial septal defect with atrial septal aneurysm. Mild tricuspid regurgitation.
12	M	6 years	CHF Cyanosis	III	Stunted growth	50%	Right atrial enlargement Rsr	DI: 54 Moderate tricuspid regurgitation. Abnormal attachment of the antero-superior leaflet. Mild mitral valvar prolapse

Abbreviations: NYHA: NewYork Heart Association Classification; DI: Displacement index; CHF: Congestive heart failure

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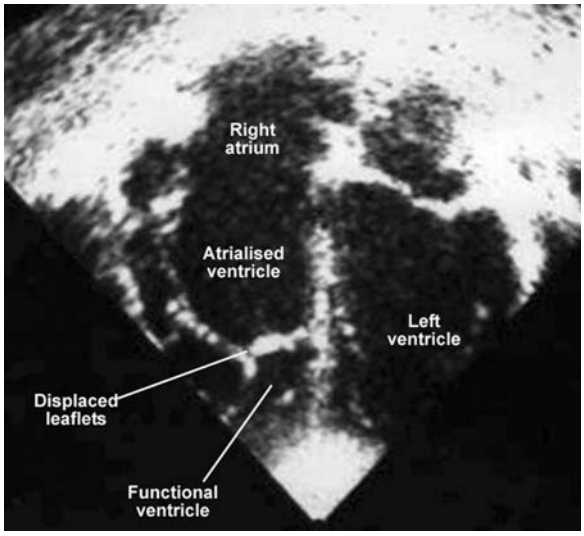

Figure 1.
The four chamber view in our ninth patient shows an index for displacement of the tricuspid valve of 43.

of adults from Lebanon reported a frequency similar to ours.[6] Our study, however, is based on referrals to our centre, and it is not possible to calculate precise incidence from such data. Our study stands in contrast, however, to the experience of Correa-Villasenor et al.,[7] who found the anomaly to be more common in white children. The criterion we used for diagnosis was the one suggested by Shiina et al.[8] The age and mode of presentation for our patients were similar to those reported in the literature, and included the full range of symptoms, from lack of any symptoms to severe cardiac failure. The mean age for our patients, at 10 years, reflects the delayed diagnosis of cardiac disease, as well as the lack of facilities for neonatal and early infant cardiac surgery, in Sudan, where tertiary paediatric cardiac services were established only in 2001. Associated diseases included delayed growth in 6 boys. In our first patient, we could explain the delay by chronic renal failure, but we could find no other explanation in four of the patients. Cardiac failure could have been the cause in our third patient, but female patients with the same degree of cardiac failure did not show any effect on their growth. The association of Ebstein's malformation with impaired hearing, as seen in our first and eighth patients, had been previously reported,[9] but as far as we are aware, the association with chronic renal failure has not previously been documented. Tuberculosis is highly prevalent in Sudan, and this may explain the association found in our second patient.

It is of note that the degree of displacement of the septal leaflet, as measured by the index for displacement created by Shiinae et al.,[8] showed no correlation either with the onset of symptoms or the degree of tricuspid regurgitation. Displacement of the valvar hinge points at the site of union of the septal and mural leaflets was a feature in 11 patients, observations which had previously been emphasized by Rusconi et al.[10] In our tenth patient, the septal leaflet had failed to achieve any delamination from the muscular ventricular septum, the diagnosis of Ebstein's malformation rather than unguarded tricuspid valvar orifice being based on the observation of the elongated whip-like shape and abnormal distal attachments of the antero-superior leaflet, observations consistent with those of Leung et al.[11] Such abnormal distal attachments of the antero-superior leaflet were also noted in six other patients, in whom there was complete lack of coaptation between this abnormally tethered leaflet and the septal leaflet. In all 6 patients, tricuspid regurgitation was severe, and they had significant symptoms.

Repair of the tricuspid valve distorted by Ebstein's malformation needs skilled surgeons. Repair, as well as replacement, carries a significant rate of morbidity and re-operation.[10] With these aspects in mind, we first tried to optimize medical treatment for our cohort, proceeding to discuss surgical options carefully with both the patients and their families.

We conclude that Ebstein's malformation is frequent in our population. Although the diagnostic echocardiographic features, as expected, are similar to those well described previously in the literature, our experience has highlighted several previously unreported associations.

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