CONGENITAL TRICUSPID VALVE DISEASE AND TESTICULAR AGENESIS: A CASE REPORT

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Abstract
This is a report of a case of congenital tricuspid valve disease presenting with heart failure and pulmonary hypertension. Cardinal clinical features include breathlessness, easy fatigability since childhood, stunted growth, cyanosis, finger clubbing, a pansystolic murmur loudest at the left sternal edge in the fourth intercostal space, and testicular agenesis. The rarity of congenital tricuspid valve disease is noted as well as the importance of a thorough physical examination of patients with congenital heart disease in order to detect possible associated non-cardiac defects.

Key words: Tricuspid valve disease, heart failure, pulmonary hypertension, testicular agenesis

Introduction
The incidence of congenital heart disease (CHD) varies between 3.3 and 10.0 per 1000 live births. Commonly reported NCD include well-recognised syndromes like Down’s syndrome, Kartagener’s syndrome, Holt-Oram syndrome and Marfan’s syndrome. Others include asplenia, anorectal malformation, oesophageal atresia and duodenal atresia. Defects of the genitourinary system have been relatively few in these reports and include undescended testes, hypospadias and ambiguous external genitalia. This is a report of a case of congenital tricuspid valve disease associated with testicular agenesis.

Case report
A 13-year-old Yoruba boy was admitted to the male medical ward because of fever, cough with purulent sputum of 2 days duration and chest pain, easy fatigability, breathlessness and weakness since his early childhood. His mother had noted bluish discolouration of the tongue and lips as well as difficulty with breathing and feeding since he was born and had taken him to many doctors in several hospitals and clinics where various medications had been given without a lasting relief. She had also observed that he had not been growing well like other siblings and that he had not been doing well in the class. However, developmental milestones had occurred in time. He is the fourth of five children and is the only child with such problems. His birth was uneventful and the mother did not report any unusual illness or event during his gestation.

Physical examination revealed a chronically ill-looking, underdeveloped boy who was severely cyanosed, breathless at rest and had finger clubbing and pitting pedal oedema involving the feet and the
lower third of the legs. He was febrile with a temperature of 38.5°C. Cardiac findings included a pulse rate of 120 per minute with a regular rhythm, a raised JVP, a displaced and heaving apex beat, a right ventricular heave, a widely heard blowing pansystolic murmur maximal at the left sternal edge in the fourth intercostal space and a loud pulmonary component of the second heart sound. There was bronchial breathing as well as coarse crackles over both lung bases. The liver was enlarged with a smooth surface and a sharp lower edge about 8cm below the right costal margin. There were no testicles in the scrotal sac or groin. The clinical impression was heart failure with pulmonary hypertension due to tricuspid valve regurgitation or a VSD complicated by pneumonia and undescended testes.

In view of the history of cardiac symptoms dating back to early life, it was thought that the cardiac lesion was most probably congenital. Results of laboratory investigations are shown in table 1. Echocardiography showed marked right atrial and right ventricular dilatation while Doppler colour flow studies showed moderate tricuspid stenosis and incompetence without any thickening of the valve or vegetations. An ultrasonogram of the abdomen and pelvis showed absence of testicular tissue in the abdomen, inguinal canal or scrotal sacs. Electrocardiogram showed sinus tachycardia, right axis deviation, right atrial hypertrophy and right ventricular hypertrophy. Sputum culture yielded a growth of streptococcus pneumoniae sensitive to penicillin among other antibiotics. The lack of the facilities precluded the measurement of arterial blood gases, intracardiac and pulmonary wedge pressures, arterial oxygen saturation or cardiac angiography.

Initial treatment included intravenous crystalline penicillin 1 mega unit 6 hourly, intravenous frusemide 20 mg daily, oral acetylsalicylic acid 75 mg daily and oral propranolol 20 mg twice daily. After the first 48 hours the crystalline penicillin was discontinued. Intramuscular procaine penicillin 1 mega unit daily was started and maintained for a further 5 days while oral frusemide 20 mg daily were substituted for the injection. Response was good. He became afebrile after 5 days of the above-mentioned treatment. The cough, chest pain, breathlessness, cyanosis, pedal oedema and hepatomegaly lessened considerably. The pansystolic murmur however remained unchanged in any way. He was discharged from the ward after the third week of admission and has been seen at the medical out-patient department on 3 visits and was noted to be doing well.

Table 1: Results of laboratory investigations

<table>
<thead>
<tr>
<th>Test</th>
<th>Result</th>
<th>Normal range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Packed cell volume</td>
<td>55%</td>
<td>36 – 52%</td>
</tr>
<tr>
<td>White cell count</td>
<td>6.4 X 10^9/L</td>
<td>3.5 – 11.0 X 10^9/L</td>
</tr>
<tr>
<td>Urea</td>
<td>3.3 mmol/L</td>
<td>2.5 – 6.5 mmol/L</td>
</tr>
<tr>
<td>Sodium</td>
<td>120 mmol/L</td>
<td>136 – 145 mmol/L</td>
</tr>
<tr>
<td>Potassium</td>
<td>4.2 mmol/L</td>
<td>3.6 – 5.2 mmol/L</td>
</tr>
<tr>
<td>Chloride</td>
<td>88 mmol/L</td>
<td>94 – 108 mmol/L</td>
</tr>
<tr>
<td>Bicarbonate</td>
<td>22 mmol/L</td>
<td>24 – 32 mmol/L</td>
</tr>
<tr>
<td>Creatinine</td>
<td>60 micromoles/L</td>
<td>9 – 126 micromoles/L</td>
</tr>
<tr>
<td>ASOT</td>
<td>130 i.u./L</td>
<td>&lt; 200 i.u./L</td>
</tr>
</tbody>
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Figure 1: Chest radiograph showing patchy consolidation of the lungs, cardiomegaly, pulmonary venous congestion and a small, encysted right- sided pleural effusion

Figure 2: Echocardiogram showing marked right atrial and right ventricular dilatation. Doppler colour flow studies showed moderate tricuspid stenosis and incompetence without any thickening of the valve or vegetations
Discussion

The clinical and echocardiographic findings of tricuspid valve disease in this patient in a clinical context that strongly suggests a congenital origin is noteworthy because of its rarity as a form of CHD. The lack of improvement of the pan systolic murmur with the successful treatment of the heart failure lends further support to the congenital origin of the lesion. The low ASOT level, lack of thickening or calcification of the tricuspid valve on echocardiography and absence of associated mitral or aortic valve disease make rheumatic tricuspid valve disease unlikely. It is worthy of note that the tricuspid stenosis found on echocardiography was not clinically obvious.

Another noteworthy feature is the finding of testicular agenesis. This is in accord with the other reports which have noted the association of non-cardiac defects (NCD) with CHD. Consequently, there is a need for the thorough physical examination of patients with CHD because the early detection and treatment of such defects can improve the overall prognosis.

Other noteworthy features include finger clubbing, cyanosis and erythrocytosis all of which are due to long standing hypoxemia caused by ventilation perfusion mismatch related to longstanding or recurrent heart failure and the chest infection. Possible complications of the erythrocytosis include strokes. In view of this, there is a need for the regular surveillance of this boy in order to prevent this. Such a regular surveillance will also help to ensure that appropriate antibiotic prophylaxis is instituted whenever there is a need for dental operations or procedures such as catheterisation that may predispose to bacterial endocarditic.

References