Bardet-Biedl Syndrome with Rheumatic Aortic Regurgitation

Sir,

Bardet-Biedl syndrome is a rare autosomal recessive genetic disorder with clinical features of retinitis pigmentosa, polydactyly, obesity, learning disabilities, hypogonadism and renal abnormalities. We present here a case of Bardet-Biedl syndrome and rheumatic aortic regurgitation complicated by infective endocarditis.

A seventeen year old boy, born of first degree consanguineous marriage, presented to us with high grade fever of 10 days duration, right ankle joint pain and poor vision. Parents had noticed night blindness since four years of age. There were neither any cardiac complaints nor any history suggestive of rheumatic fever in the past.

On examination, he was short statured, his height being 150 cm (below third percentile in height for age chart) and he weighed 40 kg (BMI was 17.7). He had hexadactyly in the right foot and a remnant of a sixth finger in the right hand adjacent to the little finger which had autoamputated at the age of 8 years. Secondary sexual characters were poorly developed. His penis and testes were of prepubertal size.

On examination, pulse rate was 96/min, regular and with a collapsing character. The blood pressure recorded was 160/40-0 mm Hg. Hill’s sign and pistol shot femorals were present. Jugular venous pulse was normal. The apical impulse was hyperdynamic and located in the left sixth intercostal space. On auscultation, a loud early diastolic murmur in the left third intercostal space and a soft systolic murmur at the apex were heard. An ejection systolic murmur was also heard in the right second intercostal space. Visual acuity was 1/6 bilaterally. Fundus examination showed arterial attenuation, cellophane maculopathy, secondary optic atrophy and bony spicules with pigmentary degeneration in the periphery of the retina, suggestive of retinitis pigmentosa. On mental status examination, he had poor recall, registration, orientation and calculation abilities (mini mental status score - 4/30).

Investigations revealed ESR 140 mm in the first hour, antistreptolysin O titre-more than 400 I U, twenty four hour urine protein estimation-2.5 gm/ day (subnephrotic proteinuria), blood urea-56 mg/dl, creatinine-1.3 mg/dl and serum testosterone level-less than 20ng/dl (normal for males 200-1600 ng/dl). Both the kidneys were normal on ultrasound. Echocardiography revealed tricuspid aortic valve with severe aortic regurgitation, mild mitral regurgitation, left ventricular hypertrophy and ejection fraction of 55%. A diagnosis of Bardet-Biedl syndrome with rheumatic fever was made. The patient was treated with aspirin and procaine penicillin for ten days, and was discharged on oral penicillin and enalapril. He was readmitted 3 weeks later with high grade fever. Echocardiography revealed an aortic valve vegetation, measuring 6mm in size. Blood cultures were sterile. Patient was treated with intravenous ceftriaxone and gentamicin for 4 weeks. He had an uneventful recovery and is on regular follow up since discharge.

Our patient had four primary features of Bardet-Biedl syndrome, namely polydactyly, retinitis pigmentosa, learning disabilities and hypogonadism. He also had functional renal abnormality in the form of proteinuria.

Bardet-Biedl syndrome has been associated with congenital heart diseases like atrial septal defect, ventricular septal defect, aortic stenosis, hypertrophy of interventricular septum and dilated cardiomyopathy.3 We relate our case of rheumatic aortic regurgitation to the high incidence of rheumatic heart disease in tropical countries. This case highlights the importance of echocardiographic evaluation of patients with Bardet-Biedl phenotype.

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REFERENCES