Bardet-Biedl Syndrome with Seizures – A Rare Association

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17 year old male, who is previously asymptomatic presented with generalised tonic clonic seizures. He was born of 2° consanguinous parents and a full term normal delivery. Developmental milestones were normal. On examination, he has central type of obesity, small testis, and loss of secondary sexual characters (Figure 1). His hands are short with broad and curved fingers (Figure 2). He has flat feet on both sides. His knee joints showed genu valgus deformity. Fundus showed pigmentary changes in retina (Figure 3). There was no evidence of spasticity, ataxia, or paraparesis. His intelligence quotient is 60. His body mass index is 30.

His investigations showed Blood urea- 50 mgs%, Serum creatinine – 1.8 mgs%, Urine albumin - ++, 24 hr urine volume- 2000 ml, 24 hr urine protein – 1.17 gm/dl, Serum FSH – 27.93 mIU/ml and Serum testosterone- 162.97 ng/dl. Audiogram was normal. USG abdomen showed increased renal cortical echoes, and altered corticomedullary differentiation. EEG revealed bilateral epileptiform activity (Figure 4). MRI brain showed normal study. Based on the above findings the diagnosis of Bardet-Biedl Syndrome (BBS) was made.

He is on treatment with antiepileptic drugs. His renal disease was managed conservatively.

BBS is primarily an autosomal recessive ciliopathy characterised by progressive retinal degeneration, obesity, mental retardation, polydactyly, delayed or underdeveloped reproductive organs, and kidney abnormalities in structure and function.¹ The disorder is genetically heterogenous, with 12 BBS genes identified till date. Neurological manifestations are uncommon in BBS patients. Rarely seizures may be associated in young children with BBS.²

This case highlights the rare association of BBS with seizures.

References


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