



## A Rare Association of Bardet-Biedl Syndrome with Autism: Case Report

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### Abstract

*Bardet-Biedl syndrome (BBS) is a rare autosomal recessive condition characterized by retinitis pigmentosa, postaxial polydactyly, obesity, mental retardation, hypogonadism and renal anomalies. Autism is rarely described in literature with BBS. Although mental retardation is generally associated with Bardet-Biedl (BBS) syndrome, autism is an unusual comorbidity.*

**Keywords:** Bardet-Biedl syndrome; Autism

### Introduction

Bardet-Biedl syndrome (BBS) is an autosomal recessive condition with a variable spectrum of clinical features. The principal manifestations are retinitis pigmentosa, postaxial polydactyly, central obesity, mental retardation, hypogonadism, and renal dysfunction<sup>1</sup>. Nearly a third of children suffering from BBS have associated behavioural problems, although these are rarely the presenting symptoms.<sup>2</sup>

### Case Report

A 13-year-old female child presented with symptoms of poor communication skills,

excessive weight gain, inability to see at night and poor learning skills. She was born of non-consanguineous marriage with no significant antenatal and postnatal history. Her BMI was 34 kg/m<sup>2</sup> and her developmental age was equivalent to 8 year. She had poor social interaction, lack of eye-to-eye contact and paucity of non verbal communication. Psychometric assessment on Indian Scale for Assessment of Autism, the score was 130 which means moderate autism.<sup>3</sup> There was postaxial polydactyly of left hand and left foot. Fundus examination revealed pallor, waxy discs with bony spicules, bilaterally attenuated vessels and retinal pigmentary changes of retinitis

pigmentosa. He had mild night blindness and a constricted peripheral visual field. suggestive of retinitis pigmentosa.

Laboratory analysis including complete blood count, urinalysis, biochemical and thyroid hormone profile, FSH, LH level as well as MRI of the brain, ultrasonography of abdomen, echocardiography and ECG were normal. Our case was labeled as BBS based on presence of 4 primary features and 4 secondary features along with autism<sup>4</sup>. Multimodality treatment with comprehensive behavior therapy was started to the patient and parental counseling was given on a regular basis.

### Discussion

Individuals with BBS frequently have behavioural disorders which include emotional disturbances, depression, thought disturbance, lack of social dominance and obsessive compulsive behavior.<sup>5</sup> There are also frequent volatile outbursts, inappropriate and disinhibited behaviour, inability to recognise social cues and shallow affect.<sup>6</sup> Internalising problems including withdrawn, somatic and anxious/depressed moods were frequent, as were problems with social behaviour, thought disturbance and attention deficit disorder.<sup>7</sup> in relative to externalizing behavior. However, BBS was rarely reported with autistic disorder in the existing literature, although isolated symptoms were described.<sup>8</sup> This case report has highlighted the importance of having a high index of suspicion in syndromic childhood obesity and emphasises the importance of detailed case history and clinical examination.

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