CASE REPORT

Autism: a rare presentation of Bardet-Biedl syndrome

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SUMMARY

Although mental retardation is generally associated with Bardet-Biedl (BBS) syndrome, a rare autosomal recessive disorder with multisystem involvement, autism is an unusual comorbidity. An 8-year-old boy presented to our psychiatry department with poor social skills and night blindness. On further assessment autism, mild mental retardation, retinitis pigmentosa, polydactyly and syndactyly, obesity, micropenis, maldescended testis, hypodontia and high-arched palate were noted and subsequently a diagnosis of BBS was made. To the best of our knowledge, this is the first reported case of BBS with autism from eastern India; it also emphasises the importance of thorough physical examination even in a patient presenting with pure psychiatric symptoms.

BACKGROUND

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive disorder involving immotile cilia,¹ characterised by retinal dystrophy, obesity, postaxial polydactyly, renal dysfunction, learning difficulties and hypogonadism.² Nearly a third of children suffering from BBS have associated behavioural problems, although these are rarely the presenting symptoms.²

CASE PRESENTATION**History**

An 8-year-old male child presented to our psychiatry outpatient department with symptoms of poor academic performance, difficulty in peer interaction and inability to see at night. His parents enrolled him in a school, but he eventually dropped out because of poor learning ability.

He was born out of consanguineous marriage (second-degree relatives). He is youngest of three siblings. His elder brother also suffered from learning difficulty and visual problems and died at the age of seven after suffering a brief illness suggestive of renal failure. Our patient's motor and developmental milestones were delayed, with initiation of walking and speech at 3 and 5 years of age, respectively.

Mental status examination revealed he had very little social interaction, lack of eye-to-eye contact and blunted affect. There was neither satisfactory development of spoken language nor other modes of communication such as gesture or mime. There was also repetitive use of some particular words and sounds. He could not participate in make-believe play appropriate to his age.

Physical examination

On physical examination, his height and weight were 120.5 cm and 41 kg, respectively, with a

resultant body mass index of 28, which indicated obesity (figure 1). His pulse was 84/min and blood pressure was 110/70 mmHg which denotes hypertension. Oral examination revealed hypodontia and a high-arched palate (figure 2). He had postaxial polydactyly of both hands (figure 3) and left foot (figures 4 and 5) with syndactyly of the right hand (figure 3). The little finger of his left hand had been surgically removed at the age of 2 months. Maldescended testes were detected on genital examination, his penis was small (stretched penile length 2.4 cm) and buried in adipose tissue (figure 6). Neurological examination showed signs of mild ataxia, poor coordination, dysarthria and proximal lower limb weakness (power grading 4/5). These findings along with the symptoms of difficulty in night vision aroused our clinical suspicion and we referred him to the ophthalmologist for funduscopy.

Figure 1 Truncal obesity.

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Figure 2 High-arched palate and hypodontia.

INVESTIGATIONS

Ophthalmological examination

Visual acuity was 6/36 in both eyes. Fundus examination showed a waxy pallor of the optic disc, bony spicules, bilaterally attenuated vessels and retinal pigmentary changes of retinitis pigmentosa. He had mild night blindness and a constricted peripheral visual field.

Laboratory investigation

Laboratory analyses including complete blood count, urinalysis, biochemical and thyroid hormone profile as well as MRI of the brain were normal. Aspartate transaminase, alanine transaminase and serum creatinine were mildly raised.

Ultrasound revealed fatty liver and increased cortigenicity of renal cortex along with irregular margins. ECG and echocardiogram were normal.

Psychometric assessment

- ▶ The patient's IQ on Malin's Intelligence Scale for Indian Children³ was 57 which indicates mild mental retardation, with 50% disability. Verbal IQ was 65 and performance IQ was 50.
- ▶ Childhood Autism Rating Scale⁴ indicated 'mild-to-moderate' symptoms of autism spectrum disorder.
- ▶ On Indian Scale for Assessment of Autism⁵ the score was 120 which means moderate autism.
- ▶ Testing was followed by detailed clinical interviews by two psychiatrists who individually confirmed a diagnosis of

Figure 3 Polydactyly and syndactyly of upper limbs.

Figure 4 Polydactyly of lower limbs.

childhood autism (F84.0, International Classification of Diseases, 10th edition).⁶

DIFFERENTIAL DIAGNOSIS

According to the modified diagnostic criteria, which were defined after a study conducted in England in 109 patients with

Figure 5 Polydactyly of left foot.

Figure 6 Micropenis and maldescended testes.

BBS,² patients who had four primary characteristics or three primary and two secondary criteria were identified as BBS (table 1).

Our case had the entire primary and three of the secondary diagnostic criteria.

TREATMENT

Individual and parental counselling was given on a regular basis. Special schooling was suggested. A comprehensive behaviour

therapy package was developed for the individual child to help with his autistic features. Regular liaison was carried out with other departments for treatment of other comorbidities.

DISCUSSION

Clinical features

BBS is an autosomal recessive ciliopathic disorder first described by Bardet-Biedl in the 1920s.⁷ It is characterised by rod-cone dystrophy (>90%), truncal obesity (72%), postaxial polydactyly (66%), cognitive impairment (62%), male hypogonadotropic hypogonadism (88%), complex female genitourinary malformations, and renal abnormalities and impairment (15–20%).^{8,9}

Other features, though not always present, are hepatic fibrosis, diabetes mellitus, reproductive abnormalities, endocrine abnormalities, short stature, developmental delay and speech deficits.² The full spectrum of clinical features is found in only 40–45% of BBS cases.¹⁰

Genetics

The diagnosis of BBS is established by clinical findings. Eighteen genes (BBS1–BBS18) are known to be associated with BBS till date.¹¹

Behavioural problems

Behavioural problems are described in about 33% of individuals with BBS which include emotional immaturity, outbursts, depression, lack of social dominance and obsessive compulsive behaviour.² There are also frequent volatile outbursts, inappropriate and disinhibited behaviour, inability to recognise social cues and shallow affect.¹² Clinically significant levels of externalising behaviour including aggression and delinquency were rarely reported. In contrast, internalising problems including withdrawn, somatic and anxious/depressed moods were frequent, as were problems with social behaviour, thought disturbance and attention deficit disorder.¹³

Although the existing literature on BBS does not report autistic disorder as a common comorbidity, studies have reported consistent occurrence of autistic signs and symptoms in these paediatric patients. In this particular case a definite diagnosis of autism was made.

Conclusion

To the best of our knowledge, this is the first case of BBS reported from eastern India.

BBS associated with autistic disorder has not been reported so far, although isolated symptoms were described in previous studies.

Careful examination and subsequent investigations revealed the rare comorbidity of BBS in a case that presented with typical features of autism and intellectual disability. This emphasises the importance of detailed history taking and physical examination even in typical psychiatric presentations.

Table 1 Diagnostic Criteria of Bardet-Biedl syndrome

Features	Our case
Primary features	
1. Rod-cone dystrophy	+
2. Polydactyly	+
3. Obesity	+
4. Learning disabilities	+
5. Hypogonadism in males	+
6. Renal anomalies	+
Secondary features	
1. Speech disorder/delay	+
2. Strabismus/cataracts/astigmatism	–
3. Brachydactyly/syndactyly	+
4. Developmental delay	+
5. Nephrogenic diabetes insipidus	–
6. Ataxia/poor coordination/imbalance	–
7. Mild spasticity	–
8. Diabetes mellitus	–
9. Dental crowding/hypodontia/small roots	+
10. Left ventricular hypertrophy/congenital	–
11. Heart disease	–
12. Hepatic fibrosis	–

Learning points

- ▶ Bardet-Biedl syndrome (BBS) is characterised by retinal dystrophy, obesity, postaxial polydactyly, renal dysfunction, learning difficulties and hypogonadism.
- ▶ Autism may be a presenting feature of BBS.
- ▶ In all psychiatric cases detailed physical examination should be routinely performed to avoid missing important comorbidities.

Contributors SSC and PG detected the case. AT and GD reviewed literature. SSC, PG and GD wrote the manuscript. AT provided inputs for the final manuscript.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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