Case report

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Selective mutism as a rare presentation of Bardet-Biedl syndrome

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Introduction

Bardet-Biedl syndrome (BBS), also known as Laurence Moon-Bardet-Biedl syndrome (LMBBS), is a rare ciliopathic autosomal recessive genetic disorder with a prevalence rate of 1 in 100000 to 160000 (1). BBS is diagnosed by Beales modified diagnostic criteria (at least four major clinical signs or three major and two minor) (2). There are six primary clinical features: retinal dystrophy, postaxial polydactyly, obesity, genital anomalies, renal anomalies, and learning disorders develop over years, and diagnosis is usually made in late childhood or early adulthood (3).

Poor vision is one of the major features of Bardet-Biedl syndrome occurring due to retinal degeneration (4). Problems with night vision become apparent by mid-childhood, followed by tunnel vision. Most BBS patients also develop blurred central vision (poor visual acuity) and become legally blind by adolescence. Abnormal weight gain typically begins in early childhood and becomes complicated by type 2 diabetes, hypertension and hypercholesterolemia (5). Other major signs and symptoms of Bardet-Biedl syndrome include polydactyly, intellectual learning disability and hypogonadism (2). Many people with Bardet-Biedl syndrome also have renal abnormalities (6). Additional features of Bardet-Biedl syndrome can include impaired speech, delayed development of motor skills such as standing and walking, behavioural problems such as emotional immaturity and inappropriate outbursts, clumsiness, and poor coordination.

Confirmation can be made by molecular genetic testing. Without family history or consanguinity, prenatal diagnosis is rarely made (7). Pathogenic or likely pathogenic variants in at least 24 genes have been associated with BBS.

A systematic study of the behavioural phenotype of LMBBS has yet to be performed (8). Notably, clinical levels of externalising behaviour, including aggression and delinquency, were rarely reported. In contrast, internalising problems, including withdrawal, somatic, and anxious/depressed mood, were frequent (8).

We report how a teenage boy diagnosed with Laurence Moon-Bardet Biedl Syndrome, coming from a poverty-stricken family and presenting with selective mutism, was managed in the child and adolescent psychiatry services. We could not find any other reports of children with selective mutism and BBS.

Case history

Master N is a 15-year-old male clinically diagnosed with Bardet-Biedl syndrome at two years of age and is on follow-up at the endocrinology and nephrology services. He presented to child and adolescent mental health services with a history of stranger avoidance since early childhood. He was reluctant to engage with and refused to make eye contact or initiate speech with unfamiliar individuals. These behaviours contrast with his typical interactions with family members, with whom he communicates using three to four-word sentences and responding appropriately to their emotions. He becomes restless when separated from his family members in unfamiliar environments.

As a result, he would stay isolated in the classroom, playground and during relative's gatherings. He does not exhibit any aggressive behaviours and as such the others would not mind his presence in spite of the lack of engagement.

There is no history of restricted repetitive behaviours or sensory hyper-hypo-sensitivities. He had no irritability or aggression towards strangers, nor were there any symptoms of hyperactivity or inattention.

N was delivered by elective lower segment caesarean section due to gestational diabetes mellitus and advanced maternal age. He had difficulties walking without support and articulating single words by the age of two years. Then, he was referred to paediatric services, where he was diagnosed with LMBBS. At 10 years of age, he had been diagnosed with intellectual developmental disorder and recommended to be followed up at a neuro developmental clinic.

Master N attended mainstream education classes until 11 years of age and discontinued schooling after the pandemic brought substantial financial constraints to the family. Currently, N can only recognise the colour red, count to three, and identify basic body parts, but he cannot recognise letters, shapes or handle money. N demonstrates independence in basic self-care and has achieved continence. However, he requires assistance with certain instrumental activities of daily living, such as buttoning shirts and tying shoelaces.

N is the youngest child and the fourth pregnancy of a non-consanguineous marriage. His father, aged 66, is a manual labourer, and his mother, aged 58, is a housewife. His mother's first pregnancy resulted in a third-trimester miscarriage. His oldest brother (age 30) is unemployed and has worked as a semi-skilled labourer. He has a 25-year-old brother diagnosed with intellectual developmental disorder, polydactyly, bilateral cataracts, and hypothyroidism.

Master N's medical history includes obesity, hypercholesterolemia managed with atorvastatin and left-sided renal parenchymal disease. He had undergone orchidopexy for undescended testes at one year of age. On examination, N had acanthosis nigricans and polydactyly in feet.

Master N was seated at the edge of the chair with downcast eyes, only engaged with his mother and had an anxious mood. He was commenced on oral escitalopram for his overwhelming anxiety, including selective mutism. We commenced an individual educational plan for Master N with specific strategies for a consistent schedule, clear expectations and visual aids. An educational therapist introduced him to hands-on activities using the multi-sensory approach. Further, tasks were broken down into smaller, more manageable steps. He will be attending the endocrinology and renal clinics as usual.

The family was registered with the divisional secretariat office to obtain a monthly allowance for managing N's financial requirements. The mother was in denial about his drawbacks and the brother's diagnosis, which was also possibly the same. We did not prioritise moving her out of the denial and instead focused on obtaining her efforts to improve the boy's quality of life. There was a significant improvement in his engagement during follow-up visits. The educational achievements still lagged, but the family complied with the educational therapy sessions. The initial treatment plan was continued, with ongoing monitoring and adjustments as needed.

Discussion

BBS is associated with a range of psychological and neuropsychiatric manifestations. Most importantly, cognitive impairment is expected, with many patients displaying intellectual functioning that falls below normal expectations, as in the reported 15-year-old boy, though not all meet the criteria for intellectual developmental disorder (9). Further, social communication deficits are prevalent and include symptoms associated with autism spectrum disorders, which are reported in a significant proportion of patients (10). Selective mutism may be mistaken for autistic symptoms in children with intellectual delay due to difficulties in thoroughly assessing their expressions (11). The reported child's need and motivation to connect with others were clear in familiar social circles.

Patients with BBS often exhibit deficits in adaptive functioning, including functional independence and attentional capacity challenges, which likely affected social interactions and acquisition of learning skills in the reported adolescent (9). Neuropsychological evaluations in persons with BBS have identified impairments in perceptual reasoning, verbal fluency, and auditory rote learning (12). Furthermore, psychiatric manifestations such as obsessive-compulsive traits, anxiety, and mood disorders have been observed, likely due to multifactorial causes, including primary neurologic issues, social discrimination, deprivation of opportunities for schooling, and sensory deficits (12). Untreated anxiety was detected in the reported child, which may have affected the development of selective mutism (13).

Overall, the psychological aspects of BBS are complex and multifaceted, involving cognitive impairments, adaptive functioning challenges, and a high prevalence of anxiety-related symptoms. These findings underscore the need for comprehensive neuropsychological and psychiatric evaluations in patients with BBS to tailor appropriate interventions and support, which is a challenge in Sri Lanka due to the minimal number of child and adolescent psychiatrists in the country (14).

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