Original Research Paper

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Paediatrics

A CASE REPORT OF BARDET -BIEDL SYNDROME :MULTISYSTEM CILIOPATHY

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ABSTRACT **Background:** Bardet-Biedl syndrome (BBS) is a rare autosomal recessive ciliopathy characterized by a spectrum of clinical features including retinal dystrophy, polydactyly, obesity, cognitive impairment, hypogonadism, and renal anomalies. Diagnosis is often delayed due to variable presentation. We report an 8-year-old female presenting with progressive visual impairment, obesity, developmental delay, and a history of polydactyly. Ophthalmological examination revealed retinitis pigmentosa. Ultrasound imaging demonstrated bilateral renal parenchymal changes. Genetic testing confirmed BBS. The child was managed with multidisciplinary supportive care and is under regular follow-up. **Conclusion:** Early recognition of the characteristic features of BBS can help initiate timely intervention, reduce complications, and improve quality of life in affected individuals.

KEYWORDS : Bardet-Biedl syndrome, retinal dystrophy, polydactyly, ciliopathy, case report

INTRODUCTION

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive disorder resulting from defects in ciliary function. It has a prevalence of 1:140,000 in the general population, but is more common in consanguineous communities. Clinical features include progressive retinal dystrophy, postaxial polydactyly, central obesity, renal abnormalities, cognitive delay, and hypogonadism. Diagnosis is clinical, aided by genetic testing when available. Management is symptomatic and multidisciplinary.

Figure 1: Child With Post Axial Polydactyly, Central Obesity, Left Foot Equinus Deformity

CASE REPORT

An 8-year-old female child was brought to our pediatric outpatient clinic with complaints of progressive night blindness and declining school performance over the past 4 years. Parents also reported excessive weight gain and poor academic interest.

She was the second child of a third-degree consanguineous marriage. Birth history was

unremarkable, with a birth weight of 3.0 kg. A history of postaxial polydactyly of both hands and foot were noted. There was no family history of similar illness.

Clinical Examination:

- Weight: 40 kg (>97th percentile)
- Height: 122 cm (25th–50th percentile)
- BMI: 26.9 kg/m²
- Vital signs: Normal
- Neurologic: Mild hypotonia, IQ : 65
- Genitalia: prepubertal normal female genitalia

Ophthalmology: Visual acuity reduced to 6/36 both eyes; fundus examination revealed retinal pigmentary degeneration consistent with retinitis pigmentosa

Diagnosis was made based on Beales diagnostic criteria:
Primary features present (≥4 required):

- Retinal dystrophy
- Postaxial polydactyly
- Central obesity
- Cognitive impairment

Secondary Features:

- Renal anomalies (echogenic kidneys)
- Learning difficulties

Genetic confirmation: whole exome sequencing
A homozygous 3' splice site variant in intron 7 of the ***BBS5***gene (chr2:g.169497624_169497625del; Depth: 39x) that affects the invariant AG acceptor splice site upstream of exon 8 was detected.

Management:

Vision: Low-vision aids, regular ophthalmologic follow-up
Nutritional support: Calorie-restricted, high-fiber diet with physical activity recommendations
Renal care: Periodic monitoring of renal function and blood pressure
Psychological and academic support: Referral to special education services
Family counseling: Genetic counseling provided to parents regarding recurrence risk and prenatal diagnosis for future pregnancies

DISCUSSION:

BBS is a genetically heterogeneous disorder with at least 26 known genes involved, most commonly BBS1, BBS10, and BBS12. Clinical diagnosis is often delayed due to its overlapping features with other syndromes like Alström syndrome and Laurence-Moon syndrome.

Renal disease is a major cause of morbidity and mortality. Retinal dystrophy usually begins as night blindness around 5–10 years of age, progressing to legal blindness by the second decade.

There is no curative treatment. Management focuses on early diagnosis, supportive therapy, and monitoring of complications.

CONCLUSION:

Bardet-Biedl syndrome should be considered in any child with visual impairment and obesity, especially if accompanied by polydactyly or developmental delay. Early diagnosis through clinical criteria and genetic confirmation enables better management and counseling.

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