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Case Report

A rare case of Bardet-Biedl syndrome in a 10 years old child with clinical and genetic correlation

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ABSTRACT

Bardet–Biedl syndrome (BBS) is a rare autosomal recessive ciliopathy characterized by multisystem involvement, including rod–cone dystrophy, polydactyly, obesity, genital anomalies, renal dysfunction, and neurodevelopmental delay. We report a 10-year-old boy who presented with night blindness, progressive visual impairment, obesity, polydactyly, acanthosis nigricans, and insulin resistance. Ophthalmological evaluation revealed reduced visual acuity with bilateral optic disc pallor. The constellation of clinical features fulfilled the diagnostic criteria for Bardet–Biedl syndrome and was subsequently confirmed by genetic testing. Early clinical recognition of BBS is crucial to ensure timely multidisciplinary management and appropriate genetic counseling. This case underscores the importance of considering syndromic etiologies in children presenting with visual impairment, obesity, and polydactyly to facilitate prompt diagnosis and intervention.

Keywords: Bardet–Biedl syndrome (BBS), Rod–cone dystrophy, Polydactyly, Obesity, Ciliopathy, Case report

INTRODUCTION

Bardet–Biedl syndrome (BBS) is a rare, genetically heterogeneous, autosomal recessive disorder first described by Georges Bardet in 1920 and Arthur Biedl in 1922.[1] It is classified as a primary ciliopathy due to its underlying defect in ciliary structure and function, affecting multiple organ systems characterized by retinal dystrophy, obesity, post-axial polydactyly, renal dysfunction, learning difficulties and hypogonadism.[1] Many associated minor features can be helpful in making a diagnosis and are important in the clinical management of BBS.

The diagnosis is based on clinical findings and can be confirmed by sequencing of known disease-causing genes in 80% of patients.[2] The syndrome typically manifests during childhood; however, due to its variable expression and multisystem nature, delayed diagnosis is common, particularly in low-resource settings. The estimated prevalence of BBS is approximately 1 in 140,000–160,000 in Europe and North America, with a higher incidence in consanguineous populations, such as the Bedouins of Kuwait and the Newfoundland population.[2] The classic triad of BBS comprises retinal dystrophy (most commonly presenting as rod–cone dystrophy), obesity, and postaxial polydactyly.

The diagnostic framework incorporates both major and minor features, reflecting the multisystem involvement characteristic of this ciliopathy.[1] The major diagnostic criteria include rod–cone dystrophy, polydactyly, obesity,

learning disabilities, hypogonadism in males, and renal anomalies.[3] Minor features include developmental delay, speech delay, diabetes mellitus, dental anomalies, and cardiac anomalies. According to the Philip L. Beales diagnostic criteria, the presence of four or more major features, or three major plus two minor features, is considered diagnostic of BBS.[1-3] (Table 1). The purpose of this case report is to highlight BBS, a rare genetic disorder with multisystem involvement, which can be diagnosed clinically by careful assessment of the patient's major and minor diagnostic criteria as described above. Genetic confirmation further supports the diagnosis and plays a crucial role in guiding up-to-date management strategies and providing appropriate genetic counseling for the patient and the family.

Table 1: Diagnostic criteria of Bardet–Biedl syndrome (BBS).

Category	Clinical features	Notes	Frequency
Major features			
	Rod–cone dystrophy (retinitis pigmentosa)	Progressive visual loss beginning in childhood; hallmark feature	93%
	Polydactyly	Usually postaxial; present at birth, All four limbs, Upper limbs only Lower limbs only	63-81% 21% 9% 21%
	Obesity	Central/truncal; typically begins in early childhood	72-92%
	Learning difficulties / developmental delay	Variable cognitive impairment	61%
	Hypogonadism (especially in males)	Delayed puberty, small genitalia	59-98%
	Renal abnormalities	Structural or functional defects; major cause of morbidity	53%
Minor features			
	Speech delay / language impairment	Common in early childhood	54-81%
	Developmental delay		50-91%
	Dental anomalies	Hypodontia, high-arched palate, malocclusion	51%
	Diabetes mellitus / glucose intolerance	Secondary to obesity and insulin resistance	6-48%
	Congenital heart defects	ASD, VSD, PDA, or pulmonary stenosis	7%
	Hepatic fibrosis/Anomalies	Less frequent	
	Anosmia/hyposmia		60%
	Short stature / facial dysmorphism	Occasionally reported	
Diagnostic threshold			
Diagnosis confirmed	When ≥ 4 major or 3 major + 2 minor features are present	Genetic testing for BBS genes provides molecular confirmation.	

CASE REPORT

A 10 years old boy, born of a consanguineous marriage, presented to the pediatric neurology clinic with complaints of night blindness and progressive visual deterioration for the past two years. He had a good appetite with excessive weight gain since early childhood.

Over the preceding year, his parents noticed gradual darkening of the skin over the neck, axillae, and elbow creases. The child also exhibited poor memory, unclear speech, learning difficulties, and stubborn behavior. Perinatal history was unremarkable with normal birth weight, and independent walking was achieved at 18 months of age.

On examination, the patient had a syndromic appearance characterized by a round face with double chin, deep-set eyes, dental crowding, obesity (weight 65 kg, >97th percentile for age; BMI 29 kg/m²), postaxial polydactyly of the left foot. Dermatological examination revealed marked acanthosis nigricans over the neck, axillae, and elbow creases with dystrophic nails. (Figure 1). Ophthalmological evaluation showed reduced visual

acuity, more pronounced in dim light with pallor of both optic disc.

Family history revealed two affected siblings: a 3 years old brother having post axial polydactyly and hexadactyly of left hand and both feet and a 4-year-old brother having dimness of vision, hypospadias, and postaxial polydactyly and hexadactyly of both hands and feet (Figure 2).

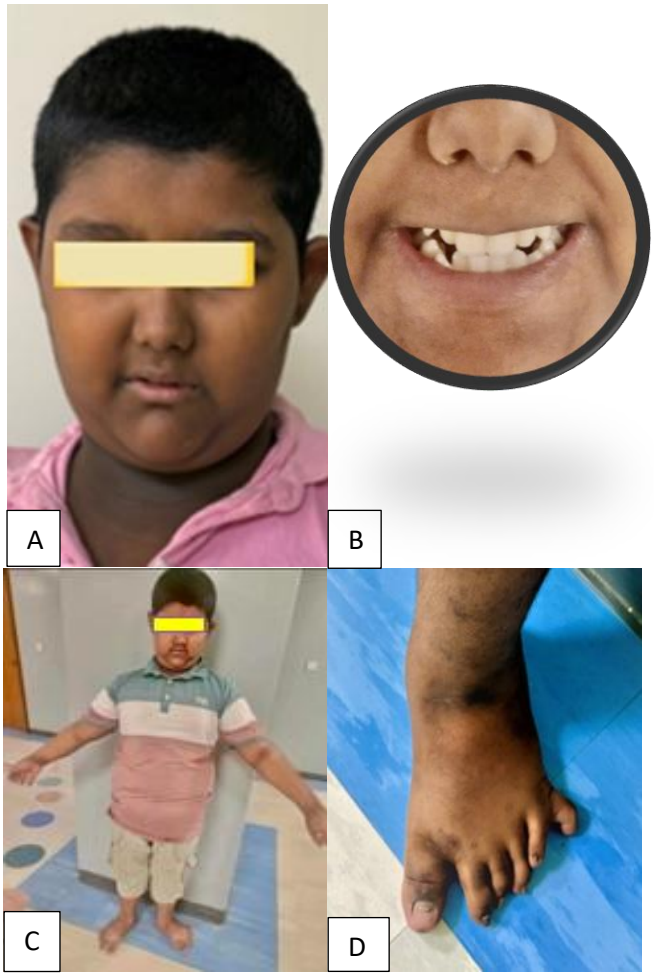

Figure 1:(A) syndromic face with marked acanthosis nigricans, (B) dental crowding, (C) obese child (BMI 29kg/m2 and (D) postaxial polydactyly and hexadactyly.

Laboratory investigations showed hemoglobin 11.9 g/dL, ESR 16 mm/hr, ALT 52 U/L, creatinine 0.6 mg/dL, random glucose 5.0 mmol/L, and HbA1c 7.8%, indicating poor glycemic control.

Thyroid function tests revealed elevated TSH (6.25 µIU/mL) with normal FT4 (1.10 ng/dL), consistent with subclinical hypothyroidism.

Renal ultrasonography demonstrated inhomogeneous parenchymal echotexture with poor corticomedullary differentiation-initiation of a renal significant change. Neurodevelopmental assessment revealed global functional delay; formal IQ testing (WISC-IV) could not be reliably completed due to poor cooperation, and functional evaluation using the Independent Behavior Assessment Scale (IBAS) showed an overall functional age of approximately 5 years.

Clinical-exome sequencing identified a pathogenic mutation in the BBS2 gene (OMIM #615981) with autosomal recessive inheritance, confirming the diagnosis of BBS type 2 (Table 2).

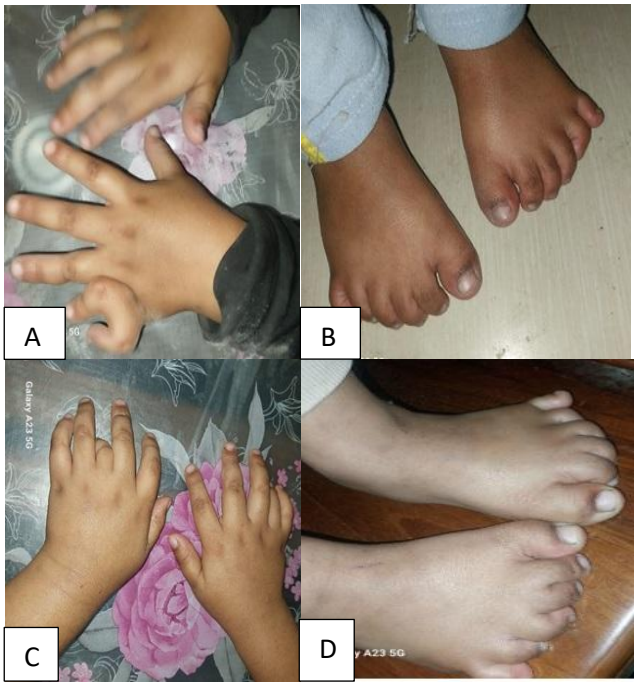

Figure 2: (A and B): 3 years old siblings having post axial polydactyly and hexadactyly of left hand and both feet and (C and D): 4 years old siblings having postaxial polydactyly and hexadactyly of both hands and feet.

Patient was managed using a multidisciplinary approach. Symptomatic and supportive treatment was initiated based on the child's clinical manifestations, with appropriate referrals to ophthalmology, endocrinology, and nephrology, as indicated. Regular developmental, visual, metabolic, and renal surveillance was advised. The parents were counseled regarding the chronic and multisystem nature of the disorder, the need for long-term follow-up, and the importance of early identification and management of associated complications. Genetic counseling was provided to explain the probable hereditary basis of the condition, recurrence risk in future pregnancies, and the benefits of genetic testing and family screening. Table 2 shows Blood test, Imaging findings, Functional Assessment by Independent Behavior Assessment Scale (IBAS) with result of Clinical-Exome sequencing. Figure 1 and 2 shows 3 years old siblings having post axial polydactyly and hexadactyly of left hand and both feet and 4 years old siblings having postaxial polydactyly and hexadactyly of hands and feet.

Table 2: Blood test reports, imaging findings, functional assessment by independent behaviour assessment scale (IBAS) and genetic test report of the patient.

Blood test reports						
Tests	**Result**			**Comments**		
Haemoglobin	11.9 gm/dl			Normal		
Glucose, (random)	5.0 mmol/l			Normal		
HBA1C:	7.8%			Poor glycaemic control		
TSH	6.25 µiu/ml - elevated			Subclinical hypothyroidism		
FT4	1.10 ng/dl - normal					
Creatinine	0.6 mg/dl			Normal		
Imaging						
Renal ultrasonography	Inhomogeneous parenchymal echotexture with poor cortico-medullary differentiation			Significant		
Functional assessment by independent behavior assessment scale (IBAS)						
Domain	Chronological age			Functional age		
Motor skills	10 years			5 years		
Socialization	10 years			5 years		
Communication	10 years			4 years		
Daily living skills	10 years			6 years		
Overall functional age	10 years			5 years		
Result of clinical-exome sequencing						
Likely pathogenic variant causative of the reported phenotype was detected						
Gene (transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
BBS2(-) (enst00000245157.11) Nm_031885.5	Exon-12	C.1465del(p.leu489trpfster58)	Homozygous	Bardet-biedl syndrome 2(omim#615981)	Autosomal Recessive	Likely pathogenic (PM2_M, PVS1_VS

Abbreviation: VS- very Strong_S - Strong_M-Moderate_Sup-Supporting

DISCUSSION

BBS is a multisystem disorder involving genetic defects in ciliary structure and function.

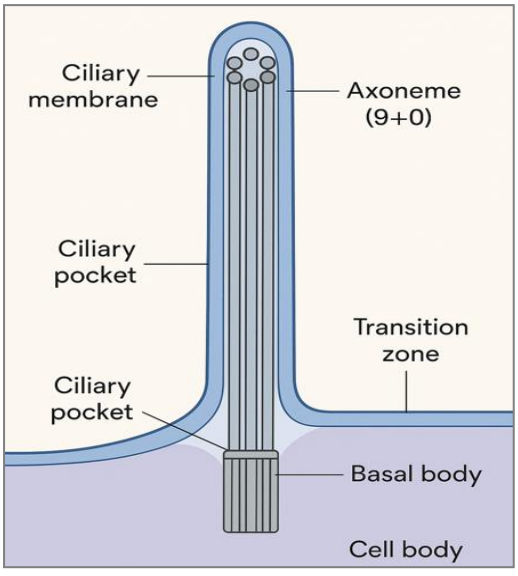

Figure 3: Structure of a typical primary cilium.

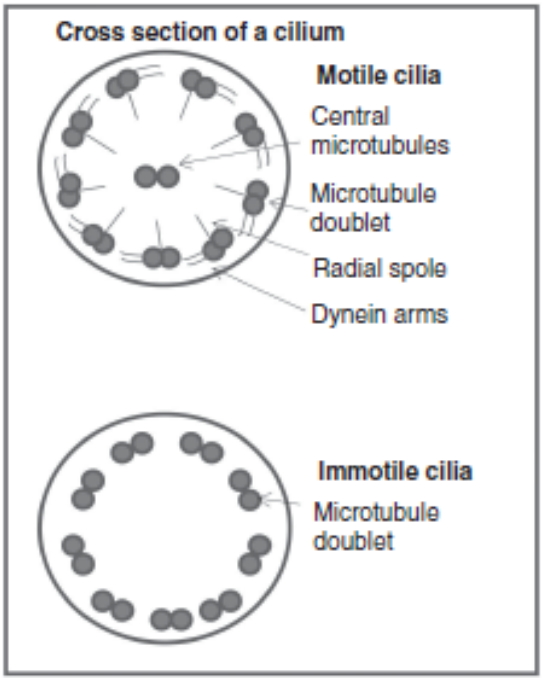

Figure 4: Motile cilia have a 9+2 microtubule pattern (with dynein arms), while primary cilia lack the central pair.

Cilia are microscopic, hair-like organelles that project from the surface of most eukaryotic cells and acts as sensory and signaling hubs. There are two major types of cilium, first one is motile cilia, actively beat to move fluid or mucus (e.g., respiratory epithelium, fallopian tubes) and second one is primary (non-motile) cilia, act as sensory antennae, crucial for signalling pathways in development and homeostasis.[3] In BBS, the primary cilia are mainly affected. Structure and function of a typical primary cilium are described below (Figure 3 and Table 3).

Table 3: Components, description and functions of a typical cilium.

Component	Description	Function
Basal body	Derived from the mother centriole	Acts as the foundation for cilium formation
Transition zone	Between basal body and axoneme	Gatekeeper — controls entry and exit of proteins
Axoneme	Core of microtubules arranged in 9+0 pattern (9 pairs around, none in the centre)	Provides structural support
Ciliary membrane	Extension of the cell membrane	Houses receptors and channels for signaling
Intraflagellar transport (IFT) system	Motor proteins (kinesin, dynein)	Transports proteins up and down the cilium

Genetic basis

BBS genes encode proteins that localize to the primary cilia and basal bodies and play a crucial role in ciliary biogenesis and function.[1]

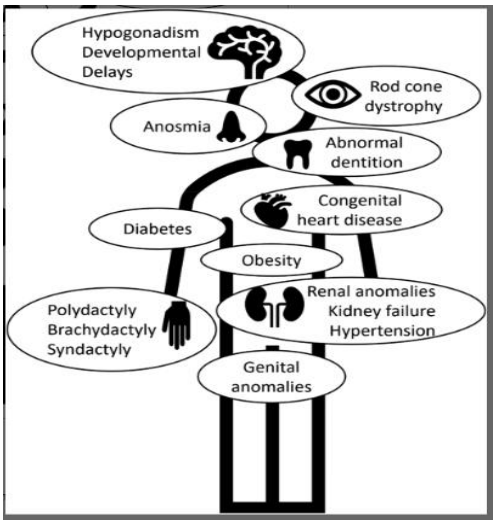

Figure: A visual representation of the multiple organ systems affected in Bardet-Biedl syndrome.

To date, more than 25 BBS-associated genes (including BBS1, BBS2, BBS10, among others) have been identified.[1,2] Pathogenic variants in these genes disrupt the BBSome complex, a multi protein assembly responsible for regulating protein trafficking into and out of the cilium and maintaining the integrity of ciliary signaling pathways. Consequently, ciliary dysfunction leads to retinal photoreceptor cilia impairment, resulting in rod–cone dystrophy and progressive vision loss; hypothalamic signaling abnormalities, leading to obesity and hormonal dysregulation; renal tubular cilia defects, causing structural and functional renal disease; and a spectrum of developmental anomalies, including postaxial polydactyly and hypogonadism. A visual representation of the multiple organ systems affected in BBS has shown in a Figure 5 by Shoemaker A.[1]

Eye

In BBS, the underlying ciliopathy results in early-onset retinal degeneration, which is the most consistent and vision-threatening manifestation of the disorder. Additional ophthalmologic features, including refractive errors, nystagmus, optic nerve abnormalities, and cataracts, may further contribute to visual impairment. However, retinal dystrophy with early macular involvement, most commonly presenting as rod–cone dystrophy, remains the hallmark of BBS-related visual loss.[9] Rod–cone dystrophy is reported in approximately 90-93% of affected individuals.[1] Clinically, patients typically present in early childhood with night blindness, followed by progressive peripheral visual field constriction, reduced color discrimination, and declining visual acuity. Night blindness usually becomes evident by 5-6 years of age, and the median age of legal blindness is around 15.5 years, although progression varies among individuals. Electroretinography (ERG) often demonstrates significant abnormalities after the age of five years, reflecting early and combined involvement of both rod and cone systems. Retinal disease is the most penetrant feature of BBS, affecting nearly all patients (100%) in some series. From a pathophysiological perspective, photoreceptors of outer segments represent specialized sensory cilia, structurally and functionally similar to primary cilia. Disruption of ciliary proteins in BBS leads to photoreceptor cilia dysfunction, resulting in progressive degeneration of rods and cones and consequent visual loss.[2]

Obesity

Obesity is a primary and highly prevalent feature of BBS, affecting approximately 72-92% of patients. Most affected individuals have a normal birth weight, but develop progressive truncal obesity during early childhood. Hyper phagia and increased food-seeking behaviour typically emerge in the first years of life, leading to rapid and often severe weight gain.[1] The pathogenesis of obesity in BBS is largely attributed to hypothalamic dysfunction, particularly involving the

melanocortin-4 receptor (MC4R) signalling pathway, secondary to defective primary cilia.[1]

Impaired ciliary function disrupts normal appetite regulation, resulting in leptin resistance and a shift toward orexigenic (appetite-stimulating) signaling, which promotes persistent hunger and early-onset obesity .[2] Under normal physiological conditions, leptin binds to its receptor on proopiomelanocortin (POMC) neurons in the hypothalamus. The protein encoded by PCSK1 cleaves POMC to generate α-melanocyte-stimulating hormone (α-MSH), which subsequently activates MC4R, leading to suppression of appetite and increased energy expenditure.[1]

Skeletal malformations

Skeletal abnormalities are a characteristic feature of Bardet–Biedl syndrome (BBS), reflecting its classification as a ciliopathy with disrupted embryonic patterning. The most consistent and clinically recognizable anomaly is postaxial polydactyly of the hands and/or feet, present at birth in the majority of affected individuals and reported in 63-81% of cases. Other commonly observed limb anomalies include brachydactyly, syndactyly (particularly between the second and third toes), shortened metacarpals or metatarsals, and a widened sandal gap. Less frequently, axial and orthopaedic abnormalities such as scoliosis, limb alignment defects, and joint problems have been described. These skeletal manifestations are attributed to impaired primary cilia–mediated signalling, particularly disruption of the Sonic Hedgehog (Shh) pathway, which plays a critical role in limb bud development and skeletal morphogenesis.[4]

Genitourinary abnormalities

BBS-related genetic mutations result in a non-motile ciliopathy, disrupting receptor trafficking and key developmental processes within the genitourinary system. Consequently, genital anomalies are common, occurring in 59-98% of patients. In males, these include hypogonadism, micropenis, and cryptorchidism, while females may exhibit vaginal atresia, urogenital sinus, uterine hypoplasia, uterine duplication, or septate vagina.[1] Renal involvement is a major contributor to morbidity and premature mortality in BBS. Structural abnormalities include vesicoureteral reflux, calyceal and parenchymal cysts, fetal lobulation, unilateral renal agenesis, and horseshoe kidney. Progressive renal dysfunction may lead to chronic kidney disease (CKD) and, in severe cases, end-stage renal disease requiring dialysis or renal transplantation.[3] The most common pathological mechanisms include chronic tubulointerstitial nephritis, cystic tubular disease, and congenital renal malformations, with additional contributions from urinary concentrating defects and glomerular involvement.[1] Metabolic comorbidities such as diabetes mellitus, hypertension, and hyperlipidemia further accelerate renal deterioration.[1] Renal ultrasonography frequently reveals fetal lobulation and loss of corticomedullary differentiation, while renal biopsy often demonstrates chronic tubule interstitial nephritis with variable fibrosis.

Neurological impairments

Neurodevelopmental involvement is common in BBS, with most affected children demonstrating mild to moderate cognitive impairment, learning difficulties, and developmental delays. Additional neurological features include speech delay, seizures, behavioral abnormalities, anosmia, hypotonia, ataxia, and poor coordination, which may contribute to delayed motor milestones. Cognitive impairment is reported in approximately 61% of cases, with variability ranging from learning difficulties to attention deficits and delayed processing speed.[1-3] Assessment of cognitive function may be confounded by early-onset visual impairment, making it difficult to distinguish primary cognitive deficits from vision-related learning challenges. Educational studies suggest that a significant proportion of children require special schooling or additional classroom support, often even before the onset of severe visual loss.[1]

Orofascial and dental anomalies

Orofacial and dentofacial anomalies are observed in more than 50% of individuals with BBS, occurring alongside other systemic features. Common extra oral manifestations include hypertelorism, strabismus, ptosis, palpebral fissure abnormalities, flat nasal bridge, retrognathia, incompetent lips, hypotonic upper lip, mouth breathing, bitemporal narrowing, brachycephaly or macrocephaly, long ears, frontal balding in males, flat nasal bridge, and maxillary atresia were reported and prominent nasolabial folds.[2] Speech disorders, often related to impaired coordination of pharyngeal and laryngeal musculature, are also common.[2] These craniofacial manifestations further contribute to the functional and psychosocial burden of the disease.

Cardiovascular anomalies

Patients with BBS may develop cardiovascular abnormalities that contribute significantly to overall morbidity. Congenital heart defects have been reported in approximately 6-10% of affected individuals and include ventricular septal defects, atrial septal defects, patent ductus arteriosus, and pulmonary stenosis. In addition, secondary cardiac manifestations, such as hypertension and cardiomyopathy, may develop as a consequence of chronic renal disease, obesity, and associated metabolic disturbances.[1]

The pathogenesis of cardiovascular involvement in BBS is thought to be related to defective primary cilia, which play a critical role in left–right axis determination during embryogenesis, leading to abnormal cardiac morphogenesis. Given these risks, early

echocardiographic assessment and long-term cardiometabolic surveillance are recommended for all patients diagnosed with BBS to facilitate early detection and prevention of progressive cardiovascular complications.[1,2]

Pathophysiology of BBS in short:

Retinal photoreceptor cilia dysfunction → rod–cone dystrophy (vision loss)

Hypothalamic signalling defects → obesity and hormonal abnormalities

Renal tubular cilia defects → structural and functional kidney disease

Developmental anomalies→ polydactyly, hypogonadism, etc.

BBS is a rare autosomal recessive ciliopathy characterized by marked clinical heterogeneity and genetic complexity.

The wide spectrum of phenotypic features—including retinal dystrophy, obesity, polydactyly, renal anomalies, and neurodevelopmental impairment—reflects pathogenic variants in multiple BBS genes involved in primary ciliary function.

Clinical–genetic correlation in BBS remains challenging due to variable expressivity, interfamilial variability, and overlapping features with other ciliopathies, underscoring the importance of integrating careful phenotypic assessment with molecular genetic testing for accurate diagnosis.

Differential diagnosis

BBS should be differentiated from other ciliopathies, obesity, retinal dystrophy syndromes/early-onset rod–cone dystrophy, postaxial polydactyly, renal structural abnormalities and neurodevelopmental involvement particularly Alström syndrome, Laurence–Moon syndrome, Cohen syndrome, and Joubert spectrum disorders.[2] (Table 4).

Table 4: Comparison table of Bardet-Biedl syndrome with its major differentials.

Feature	BardetBiedl syndrome (BBS)	Alstrom syndrome	Laurence–moon syndrome	Cohen syndrome	Joubert syndrome
Genetic group	Ciliopathy	Ciliopathy	Ciliopathy	Ciliopathy	Ciliopathy
Retinal dystrophy	Rod–cone dystrophy (early)	Cone–rod dystrophy	Present	Present	Present
Night blindness	Early	Early	Early	Variable	Variable
Polydactyly	Postaxial common	Absent	Absent	Absent	May be present
Obesity	Early-onset	Childhood	Variable	Truncal	Variable
Neurodevelopment	Learning disability	Normal–mild delay	Intellectual disability	Intellectual disability	Developmental delay
Hearing loss	Absent	Common	Rare	Absent	Rare
Renal involvement	Common	Rare	Rare	Mild	Common
Hypogonadism	Common	Common	Common	Common	Variable
Other key features	Acanthosi, dental anomalies	Cardiomyopathy	Spastic paraplegia	Neutropenia microcephaly	Molar tooth sign (MRI)
Life expectancy	Reduced	Reduced	Reduced	Near normal	Variable

Management

Management of BBS is multidisciplinary, lifelong, and supportive, aimed at early detection and prevention of complications. This Care done by the following experts are Pediatrician/Physician: Overall coordination and long-term follow-up, Pediatric Neurologist/ Developmental Specialist: Developmental delay, learning difficulties, behavioral issues, Ophthalmologist: Regular monitoring for rod–cone dystrophy and visual rehabilitation, Endocrinologist: Obesity, insulin resistance, diabetes mellitus, dyslipidemia, hypothyroidism, Nephrologist: Renal structural and functional abnormalities, Orthopedic/Plastic Surgeon: Management of polydactyly and limb anomalies and Clinical Geneticist: Diagnosis confirmation and family counseling. System-Specific Management like vision is to be done by regular ophthalmologic follow-up, low-vision aids, visual rehabilitation. Obesity and metabolic issues by dietary counseling, lifestyle modification,

physical activity, management of diabetes and dyslipidemia. Renal involvement monitored by periodic renal function tests, blood pressure monitoring and renal imaging.

Neurodevelopment issues should be managed by early intervention, speech therapy, occupational and special education support. Genital anomalies/ hypogonadism is to be managed by endocrine evaluation and appropriate hormonal management if indicated. Surveillance and follow-up done by Growth and BMI monitoring, annual ophthalmologic assessment, regular renal and metabolic screening, developmental and behavioral assessments.

Genetic Counseling should be done by counseling regarding autosomal recessive inheritance, explanation of recurrence risk (25%) in future pregnancies, advice on family screening and availability of genetic testing and discussion of prenatal diagnostic options where feasible.

CONCLUSION

Early diagnosis of BBS and a coordinated multidisciplinary approach are crucial to reduce morbidity and improve quality of life. Regular surveillance, timely management of multisystem involvement, and comprehensive genetic counseling play a pivotal role in optimizing long-term outcomes for affected individuals and their families.

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