e-ISSN: 0976-822X, p-ISSN:2961-6042

Available online on http://www.ijcpr.com/

International Journal of Current Pharmaceutical Review and Research 2025; 17(12); 2474-2482

Case Series

Unraveling Bardet–Biedl Syndrome: A Case Series of Pediatric Patients with a Multifaceted Ciliopathy

Devendra Yadav[1], Sourabh Piparsania[2], Stuti Gagrani[3]

[1]Ex-resident, Department of Pediatrics, Index Medical College, Indore, MP, India

[2]Professor, Department of Pediatrics, Index Medical College, Indore, MP, India

[3]Assistant Professor, Department of Pediatrics, Index Medical College, Indore, MP, India

Received: 10-10-2025 / Revised: 15-10-2025 / Accepted: 21-11-2025

Corresponding author: Dr. Devendra Yadav

Conflict of interest: Nil

Abstract

Background: Bardet–Biedl Syndrome (BBS) is a rare autosomal recessive ciliopathy characterized by progressive vision loss, polydactyly, obesity, intellectual disability, metabolic disturbances, and renal abnormalities. Despite its significant impact on morbidity and mortality, BBS often remains underdiagnosed, particularly in resource-limited settings where genetic testing is inaccessible.

Case Presentation: We present a case series of three pediatric patients with Bardet–Biedl syndrome (BBS) from Central India, comprising two 9-year-old female patients and one 10-year-old male patient. All three patients exhibited hallmark features of the syndrome, including progressive visual impairment due to retinitis pigmentosa, post-axial polydactyly, central obesity, and metabolic abnormalities.

Case 1 demonstrated hypoplastic ovaries with associated hormonal imbalances, while Case 2 exhibited moderate intellectual disability, hepatosplenomegaly, and insulin resistance. Case 3 presented with learning difficulties, hypogonadism, insulin resistance, dyslipidemia, and early renal parenchymal involvement, along with mild cardiac changes.

Diagnosis in all cases was established based on characteristic clinical features supported by biochemical and radiological findings, as genetic testing was not readily available. Genetic counseling was recommended for all families. A multidisciplinary management approach, including ophthalmologic surveillance, metabolic control, dietary and lifestyle interventions, endocrinological evaluation, renal monitoring, and neurodevelopmental support, was initiated to improve quality of life and delay disease progression.

Conclusion: Early recognition of BBS is critical for timely intervention and reducing complications, particularly renal failure, which is the leading cause of morbidity and mortality in affected individuals. Given the limited availability of genetic testing, clinical diagnosis based on characteristic features remains essential in low-resource settings. Increased awareness among healthcare professionals can facilitate prompt identification, allowing for comprehensive, multidisciplinary management to optimize patient outcomes.

Keywords: Bardet–Biedl Syndrome; Case Series; Ciliopathy; Retinitis Pigmentosa; Post-Axial Polydactyly; Childhood Obesity; Insulin Resistance; Renal Involvement; Intellectual Disability.

Introduction

Bardet–Biedl syndrome (BBS) is a rare inherited disorder resulting from defects in primary cilia function [1-3]. This ciliopathy is characterized by rod–cone dystrophy, kidney abnormalities, extra digits (polydactyly), cognitive impairment, central obesity, and underdeveloped reproductive organs [4]. The syndrome was first described by Bardet in 1920, followed by Biedl in 1922 [5, 6].

Its prevalence is estimated at 1 in 100,000 individuals in Europe and North America [7], while its incidence is significantly higher in the Faroe Islands (1 in 3,700) and Kuwait (1 in 17,000 live births) [8]. However, the prevalence and incidence in Sudan remain unknown.

So far, 21 genes (BBS1–BBS21) have been identified as being associated with BBS. Due to genetic diversity and the high cost of genetic testing,

these analyses are typically limited to complex cases or research purposes.

According to the diagnostic criteria proposed by Beales et al., a BBS diagnosis requires either the presence of four primary features or a combination of three primary and two secondary features [6]

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

Primary features	Secondary features
Truncal obesity	Strabismus/cataract/astigmatism
Retinitis pigmentosa/retinal dystrophy	Speech disorders/delay
Polydactyly	Developmental delay
Learning disabilities	Brachydactyly/syndactyly
Renal malformations	Behavioral disorders
Genital abnormalities (female) Hypogonadism (male)	Diabetes mellitus
	Polyuria/polydipsia (diabetes insipidus)
	Left ventricular hypertrophy (LVH) Congenital cardiac abnormalities
	Hepatic fibrosis
	Anosmia
	Craniofacial dysmorphic
	Dental crowding/high-arched palate/hypodontia/small roots
	Hirschsprung disease
	Ataxia/poor coordination

In many resource-limited healthcare settings, genetic testing remains inaccessible, making early detection difficult. The syndrome's rarity and gradual progression further complicate timely diagnosis. A delayed diagnosis can increase morbidity and mortality, with kidney impairment being the leading cause of severe health complications. Early identification is crucial to slowing disease progression. The management of BBS is supportive and requires a multidisciplinary team approach, with genetic counseling playing a critical role in family care and planning. Here, we present two cases of Bardet–Biedl syndrome, which, to the best of our knowledge, are the first documented cases from Central India, emphasizing the importance of early recognition, diagnosis, and multidisciplinary management.

Case Presentation

Case 1: We report the case of a 9-year-old girl who presented with a five-year history of progressive vision loss, gradually interfering with her daily activities. She did not report associated symptoms such as headache, dizziness, vomiting, night blindness, or color blindness. Additionally, for the past 18 months, she experienced dry skin and cracking of the palms and soles, along with severe dandruff accompanied by mild itching but without redness, rash, bleeding, localized warmth, or tenderness. On physical examination, she exhibited polydactyly in her right hand and both feet, with no other significant dysmorphic features.

Laboratory investigations revealed dyslipidemia, with increased triglycerides (26 mg/dL) and decreased HDL (34.2 mg/dL), while cholesterol (126 mg/dL), LDL (39.4 mg/dL), and VLDL (52.4 mg/dL) were within acceptable limits. Her fasting blood glucose, HbA1c, liver function tests (LFTs), and renal function tests (RFTs) were all within normal ranges. A cardiac evaluation showed sinus arrhythmia on ECG, though echocardiography was normal. Endocrine studies indicated normal prolactin and TSH levels, but reduced follicle-stimulating hormone (FSH) and luteinizing hormone (LH), with an elevated estradiol level. Imaging studies, including ultrasound and MRI of the abdomen and pelvis, confirmed hypoplastic ovaries and uterus, while an MRI of the brain was unremarkable. Bone age assessment via X-ray suggested a normal to mildly increased bone age (8–12 years). [Table 2]

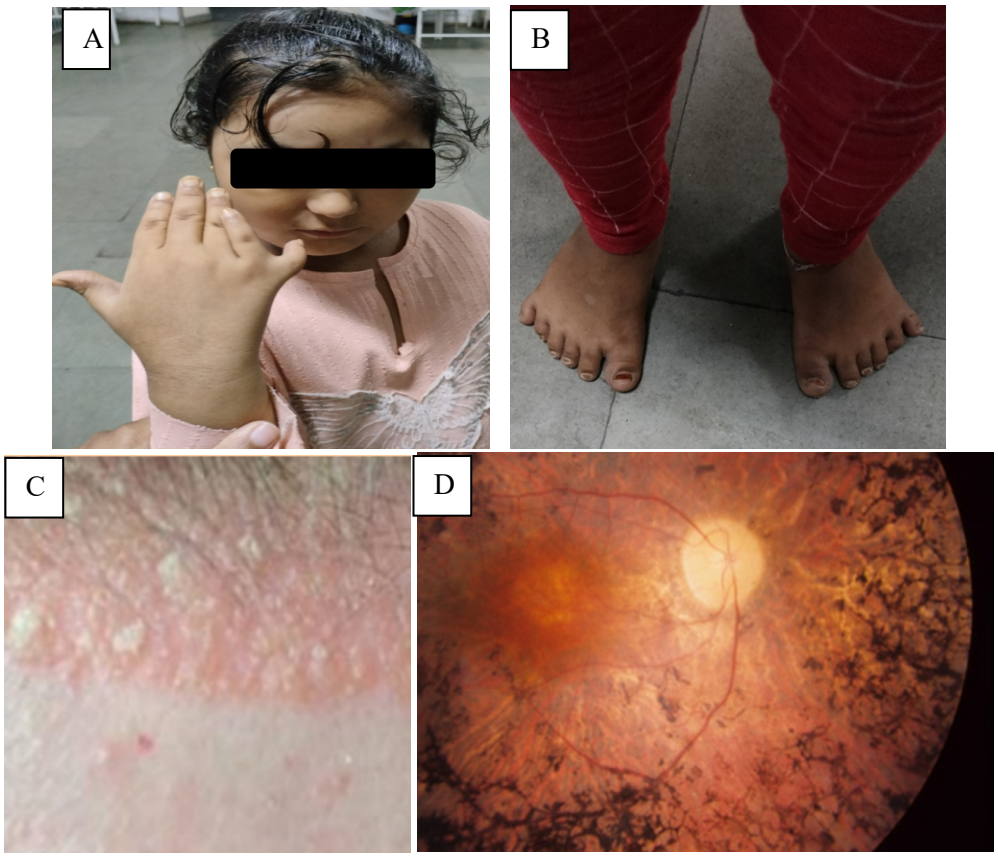

Figure 1: Clinical Presentation of Case showing; A Right upper limb post-axial polydactyly. B Bilateral lower limbs post-axial polydactyly. C. Dermatological examination revealed keratosis pilaris and seborrheic dermatitis D. Funduscopic pictures of both eyes showing retinitis pigmentosa and optic atropy

Table 2: Laboratory and Imaging Findings of the Case 1

Investigation	**Case 1**
Lipid Profile	
Cholesterol	126 mg/dL
Triglyceride	26 mg/dL
HDL	34.2 mg/dL
LDL	39.4 mg/dL
VLDL	52.4 mg/dL
Blood Investigations	
Fasting Blood Sugar	Normal
HbA1c	Normal
Liver Function Tests (LFTs)	Normal
Renal Function Tests (RFTs)	Normal
Endocrine Profile	
Prolactin	Normal
Thyroid-Stimulating Hormone (TSH)	Normal
Follicle-Stimulating Hormone (FSH)	Decreased
Luteinizing Hormone (LH)	Decreased
Estradiol	Increased
Cardiac Investigations	
ECG	Sinus Arrhythmia
Echocardiography (ECHO)	Normal
Imaging Studies	
Abdominal & Pelvic Ultrasound	Hypoplastic ovaries and uterus
MRI Abdomen & Pelvis	Hypoplastic ovaries and uterus
MRI Brain	Normal
X-ray (Bone Age)	Normal to increased (8–12 years)

Specialist consultations provided further insight into her condition. A dermatological evaluation diagnosed keratosis pilaris and seborrheic dermatitis, whereas an ophthalmological assessment revealed peripheral visual field loss. (Figure 1C) Fundoscopic examination showed findings consistent with retinitis pigmentosa. (Figure 1D) Based on these clinical, biochemical, and imaging findings, a diagnosis of Bardet-Biedl Syndrome was established.

The patient was managed with a multidisciplinary approach. To address her visual impairment, she was prescribed corrective lenses, along with Vitamin A supplementation to support retinal function. For her dermatological issues, she was advised to use emollients and moisturizers, along with ketoconazole and salicylic acid shampoo for seborrheic dermatitis. Given her metabolic concerns, dietary modifications were recommended, including a low-calorie, high-fiber diet with moderate protein intake and reduced simple carbohydrates. She was encouraged to engage in regular moderate exercise.

Recognizing the genetic nature of BBS, her parents were counseled on the importance of genetic testing and family planning. They were educated on the progressive nature of visual impairment in BBS and the need for early training in Braille and voice recognition techniques to enhance adaptability. Occupational therapy was recommended to aid in skill development and independence. With early diagnosis and comprehensive management, the goal was to optimize her quality of life and mitigate disease progression.

Case 2:

A 9-year-old female, presented with concerns of increased weight since the age of two, diminished vision since the age of five, and recurrent headaches. The parents reported that her weight gain was insidious and progressively worsened compared to children of her age. At four years old, she was found to have hepatosplenomegaly and lipid profile abnormalities, but treatment yielded no significant improvement. By the age of five, she developed visual impairment and difficulty walking, leading to a diagnosis of retinitis pigmentosa and suspected Bardet-Biedl syndrome (BBS) at another medical facility. Over the past year, she also experienced moderate to severe frontal headaches without associated symptoms like nausea, vomiting, photophobia, or phonophobia.

Her past medical history was significant for epilepsy, with no other notable illnesses reported. On physical examination, she was found to be obese, with a weight of 41 kg (>97th percentile) and a BMI of 25.25 kg/m². Her height was 132 cm (10–25th percentile), and blood pressure was measured at 108/68 mmHg. Clinical findings included facial puffiness and polydactyly. Systemic examination revealed normal cardiovascular and respiratory findings, though hepatomegaly was present.

Diagnostic investigations were conducted to further assess her condition. Echocardiography showed normal cardiac structure and function with a left ventricular ejection fraction (LVEF) of 64%. A psychological assessment revealed moderate delays in adaptive functioning, with a Social Quotient (SQ) and Developmental Quotient (DQ) suggesting moderate developmental delay. Her Intelligence Quotient (IQ) was in the range of moderate intellectual disability. Thyroid function tests were within normal limits, while fasting blood glucose was slightly low at 68.4 mg/dL. Her HbA1c was 5.8%, indicating a normal glycemic status, but fasting insulin was markedly elevated at 39.56 µU/mL. Her lipid profile showed significantly elevated total cholesterol (219.98 mg/dL) and triglycerides (364.57 mg/dL), with an LDL-C level of 105.44 mg/dL and an HDL-C of 45.62 mg/dL. Ultrasonography revealed mild hepatosplenomegaly, while her chest X-ray and complete blood count were unremarkable. [Table 3]

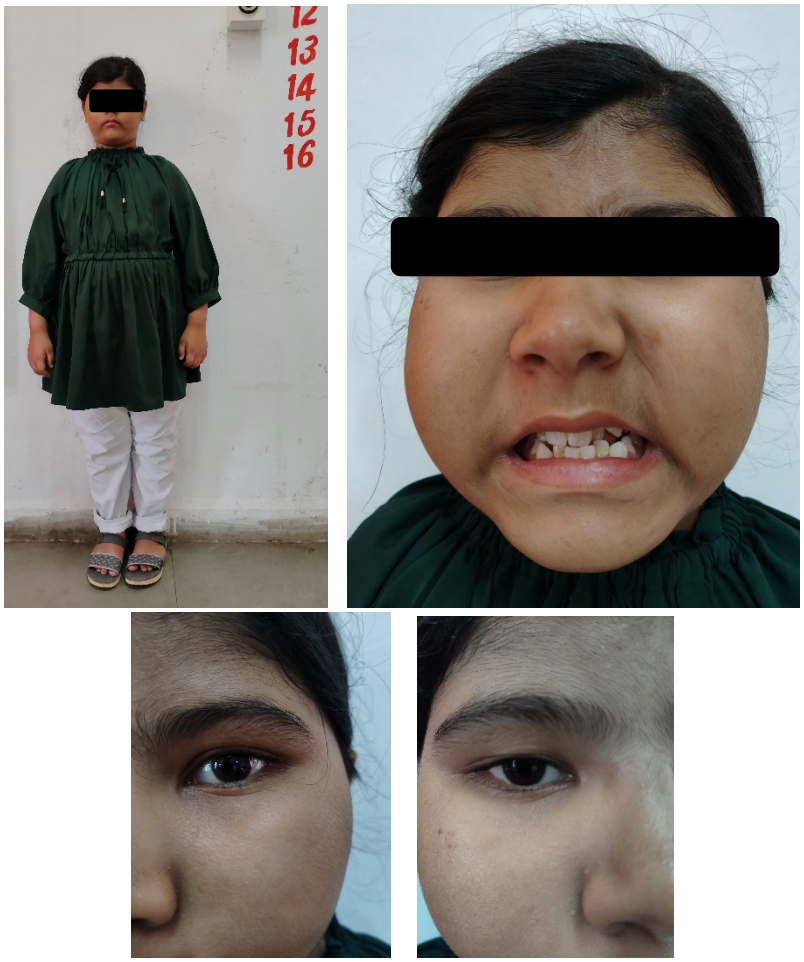

Figure 2: Clinical Presentation of Case 2 showing facial puffiness, dental crowding and polydactyly and retinal dystrophy (retinitis pigmentosa)

Table 3: Laboratory and Imaging Findings of the Case 2

Investigation	Case 2
Fasting Blood Glucose	68.4 mg/dL
HbA1c	5.8%
Fasting Insulin	39.56 µU/mL
Total Cholesterol	219.98 mg/dL
Triglycerides	364.57 mg/dL
HDL-C	45.62 mg/dL
LDL-C	105.44 mg/dL
Thyroid Function Tests	Within normal limits
Ultrasonography	Mild hepatosplenomegaly
Chest X-ray	Normal
Echocardiography	LVEF: 64%
Complete Blood Count	Within normal limits
Social Quotient (SQ)	Moderate delay
Developmental Quotient (DQ)	Moderate delay
Intelligence Quotient (IQ)	Moderate intellectual disability

Based on her clinical presentation, the provisional diagnosis was suspected Bardet-Biedl Syndrome. The patient exhibited multiple characteristic features of BBS, including early-onset obesity, retinal dystrophy (retinitis pigmentosa), cognitive impairment, polydactyly, and hepatic involvement. Her metabolic disturbances, such as elevated insulin levels and dyslipidemia, further supported the diagnosis.

A multidisciplinary management approach was recommended. Genetic counseling and testing were advised to confirm the diagnosis. Ophthalmological

follow-up was essential for monitoring retinal dystrophy progression, while endocrinological evaluation was necessary to address metabolic abnormalities. A comprehensive nutritional counseling and weight management program was suggested to mitigate obesity-related complications. Additionally, neurodevelopmental therapy and special education support were recommended to address her cognitive impairments. Regular monitoring of renal function was also advised, given the risk of renal involvement in BBS. Psychological support for the patient and her family was emphasized, along with a potential referral to a specialized BBS clinic if available.

Case 3

A 10-year-old male child presented with complaints of progressive visual impairment since the age of six years, excessive weight gain since early childhood, and delayed academic performance compared to peers. The parents also reported increased thirst and frequent urination over the past one year. There was no history of seizures, recurrent infections, or hearing loss. The child was born of a non-consanguineous marriage with no significant antenatal or perinatal complications. Family history was unremarkable for similar illnesses.

On physical examination, the child was obese with a weight of 45 kg (>97th percentile) and a BMI of 26.8 kg/m². His height was 135 cm (25th percentile). Post-axial polydactyly was noted in both feet, while upper limbs were normal. Facial features revealed mild dysmorphism with dental crowding. Genital examination showed micropenis with small testes suggestive of hypogonadism. Blood pressure was 112/70 mmHg. Systemic examination was otherwise unremarkable.

Ophthalmological evaluation revealed reduced visual acuity with peripheral visual field constriction. Fundoscopic examination showed characteristic features of retinal dystrophy consistent with retinitis pigmentosa. Neurodevelopmental assessment indicated mild to moderate learning difficulties, with poor scholastic performance.

Laboratory investigations showed impaired fasting glucose (112 mg/dL) with an HbA1c of 6.1%, suggestive of prediabetes. Fasting insulin levels were elevated (31.8 μU/mL), indicating insulin resistance. Lipid profile revealed elevated triglycerides (298 mg/dL) and total cholesterol (208 mg/dL), with reduced HDL cholesterol (38 mg/dL). Renal function tests were within normal limits. Thyroid function tests were normal.

Ultrasonography of the abdomen revealed increased echogenicity of both kidneys suggestive of early renal parenchymal involvement, without evidence of hydronephrosis. Echocardiography showed mild concentric left ventricular hypertrophy with preserved systolic function. MRI brain was normal.

Based on the presence of multiple primary features—retinal dystrophy, post-axial polydactyly, central obesity, learning difficulties, hypogonadism—and secondary features such as insulin resistance and renal involvement, a clinical diagnosis of Bardet–Biedl syndrome was established.

The patient was managed with a multidisciplinary approach. Dietary counseling and lifestyle modification were initiated to address obesity and metabolic abnormalities. Regular ophthalmological follow-up was advised to monitor visual deterioration. Endocrinology consultation was sought for management of insulin resistance and hypogonadism. Nephrology follow-up was planned for periodic assessment of renal function. Genetic counseling was provided to the family, emphasizing the hereditary nature of the disorder and the importance of long-term follow-up.

Discussion

Bardet–Biedl syndrome (BBS) is a rare genetic disorder affecting multiple systems. It follows an autosomal recessive inheritance pattern. In North American and European populations, the estimated incidence is approximately 1 in 150,000–160,000, though this rate is higher in regions with frequent consanguineous marriages [5].

Over the last decade, the loss or dysfunction of cilia has been associated with several human disorders collectively referred to as ciliopathies. BBS is classified as a ciliopathy, as it disrupts the function of multiple ciliated organs, leading to systemic symptoms.

BBSomes are proteins encoded by Bardet–Biedl genes that facilitate primary cilia biogenesis and function.

To date, 21 BBS genes (BBS1–BBS21) have been identified, with BBS1 and BBS10 being the most prevalent, accounting for over 20% of cases. Mutations in BBS1 typically present later than those

in BBS10 due to a milder phenotype and delayed onset of retinal degeneration. Given the high cost and time-consuming nature of genetic testing, it is usually reserved for complex cases and research. BBS can often be diagnosed based on clinical criteria alone.

The primary clinical characteristics of BBS include rod–cone dystrophy, post-axial polydactyly, central obesity, cognitive impairment, hypogonadism, complex genital abnormalities, and renal dysfunction [9]. Secondary symptoms involve speech delays, ocular abnormalities (such as strabismus, cataracts, and astigmatism), brachydactyly or syndactyly, developmental delays, ataxia, diabetes mellitus, craniofacial anomalies, nephrogenic diabetes insipidus, hepatic fibrosis, and left ventricular hypertrophy/congenital heart defects [6, 10].

A diagnosis of BBS is typically confirmed when a patient presents with either four primary symptoms or three primary symptoms alongside two secondary symptoms. According to a large UK-based study, the average age of diagnosis is 9 years.

Various renal abnormalities have been documented in BBS, including chronic kidney disease (CKD), parenchymal cysts, calyceal clubbing, fetal lobulation, renal scarring, unilateral agenesis, dysplastic kidneys, renal calculi, and vesicoureteric reflux [11]. The progression of renal disease in BBS remains uncertain. Renal impairment may arise due to primary causes, such as cystic kidney disease, or secondary factors, including hypertension, diabetes, or metabolic syndrome. Kidney failure is the leading cause of mortality among individuals with BBS, accounting for 25% of deaths by age 44 [12]. Management of renal failure in BBS patients follows standard protocols, including chronic peritoneal dialysis, hemodialysis, and kidney transplantation [13, 14].

Studies indicate that adults with BBS have a higher prevalence of insulin resistance and metabolic syndrome compared to the general population, contributing to increased cardiovascular mortality. However, many individuals have not yet developed overt diabetes, offering an opportunity for early intervention through lifestyle modifications [15]. The loss of BBS genes, specifically BBS1 or BBS4, has been linked to a significant increase in pancreatic β-cell production.

To identify BBS patients, medical history, family history, physical examination, laboratory tests, and genetic testing are essential components of the evaluation strategy. A BBS diagnosis should be considered in individuals exhibiting any of the major features outlined in Table 1. Identifying these characteristics should prompt further evaluation to confirm the diagnosis.

Additionally, BBS should be suspected in fetuses or infants presenting with structural kidney abnormalities, genitourinary malformations, or polydactyly, as these may be early indicators of the condition. Central obesity, an early and notable feature, often becomes apparent within the first year of life. Features of cone–rod dystrophy (such as photophobia, decreased visual acuity, and impaired color discrimination) and CKD (manifesting as polyuria and polydipsia) generally emerge by school age, whereas signs of hypogonadism (such as delayed puberty) may not become evident until adolescence. Currently, no curative treatment exists for BBS, emphasizing the importance of early diagnosis and routine follow-up assessments [4, 6].

Managing BBS requires a multidisciplinary team, including pediatricians, nephrologists, orthopedic surgeons, cardiologists, ophthalmologists, dental specialists, speech pathologists, and audiologists. Given the certainty of blindness associated with cone–rod dystrophy, early educational planning is essential. Supportive measures such as Braille, mobility training, adaptive living skills, computing skills (e.g., voice recognition and transcription software), and large-print reading materials can assist patients while vision remains intact [13, 14].

To address obesity, a low-carbohydrate diet and regular aerobic exercise, such as walking or cycling with necessary adaptations for blindness, are recommended. Having a support companion can help maintain a healthy lifestyle. However, in resource-limited areas, the absence of trained dietitians and blindness-related challenges complicate diet management for BBS patients. The metabolic syndrome and obesity-related complications in BBS should be treated similarly to the general population. For cosmetic purposes, surgical removal of extra digits can be considered [16].

Individuals with anosmia (impaired sense of smell) should have alternative methods for detecting potential hazards, such as spoiled food or smoke. Learning disabilities may necessitate enrollment in

specialized educational programs. Lastly, genetic counseling is highly beneficial for affected individuals and their families [13, 14].

Conclusion

Early recognition of Bardet–Biedl syndrome (BBS) is crucial for timely intervention, lifestyle modifications, and preventive measures to minimize complications and improve the patient's quality of life. Given its rarity, BBS often goes unrecognized by clinicians, leading to delayed diagnosis and management. Renal failure remains the primary cause of morbidity and mortality in affected individuals, underscoring the need for early detection to slow disease progression. A combination of post-axial polydactyly, vision impairment, cognitive disabilities, renal abnormalities, and obesity should raise suspicion for BBS, prompting further evaluation. Multidisciplinary care and proactive management play a pivotal role in enhancing long-term outcomes for individuals living with BBS.

References

1. Elawad, O.A.M.A., Dafallah, M.A., Ahmed, M.M.M. *et al.* Bardet–Biedl syndrome: a case series. *J Med Case Reports* 16, 169 (2022). https://doi.org/10.1186/s13256-022-03396-6
2. Ansley SJ, Badano JL, Blacque OE, Hill J, Hoskins BE, Leitch CC, Kim JC, Ross AJ, Eichers ER, Teslovich TM, Mah AK, Johnsen RC, Cavender JC, Lewis RA, Leroux MR, Beales PL, Katsanis N. Basal body dysfunction is an expected cause of pleiotropic Bardet–Biedl syndrome. Nature. 2003;425:628–33. https://doi.org/10.1038/nature02030.
3. Adams M, Smith UM, Logan CV, Johnson CA. Recent advances in the molecular pathology, cell biology and genetics of ciliopathies. J Med Genet. 2008;45:257–67. https://doi.org/10.1136/jmg.2007.054999.
4. Bardet G. Sur un syndrome d'obesitecongenitale avec Polydactilieetretinitepigmentaire (contribution a l'etude des formescliniquesdel'obesitehy pophysaire) [On congental obesity syndrome with poly-dactyly and pigmentary retinitis (contributions to the study of clinical forms of pituitary obesity)]. Amedee Le Grand. 1920;470:407.
5. Beales PL, Elcioglu N. New criteria for improved diagnosis of Bardet–Biedl syndrome: results of a population survey. J Med Genet. 1999;36:437–46.
6. Biedl A. Ein Geschwisterpaar mit adiposo-genitaler Dystrophie [A pair of siblings with adiposogenital dystrophy]. Dtsch Med Woschenschr. 1922;48:1630.
7. Forsythe E, Beales PL. Bardet–Biedl syndrome. Eur J Hum Genet. 2013;21(1):8. https://doi.org/10.1038/ejhg.2012.115.
8. Hjortshøj TD, Grønskov K, Brøndum-Nielsen K, Rosenberg T. A novel founder BBS1 mutation explains a unique high prevalence of Bardet– Biedl syndrome in the Faroe Islands. Br J Ophthalmol. 2008;93(3):409–13.
9. Green JS, Parfrey PS, Harnett JD. The cardinal manifestations of Bardet– Biedl syndrome, a form of Laurence–Moon–Biedl syndrome. N Engl J Med. 1989;321(15):1002–9.
10. Moore SJ, Green JS, Fan Y. Clinical and genetic epidemiology of Bardet– Biedl syndrome in Newfoundland and Labrador: a 22-year prospective, population-based, cohort study. Am J Med Genet A. 2005;132(4):352–60.
11. Beales PL, Elcioglu N, Woolf AS, Parker D, Flinter FA. New criteria for improved diagnosis of Bardet–Biedl syndrome: results of a population survey. J Med Genet. 1999;36(6):437–46.
12. Forsyth R, Gunay-Aygun M, et al. Bardet–Biedl syndrome overview. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Mirzaa G, et al., editors. GeneReviews®. Seattle: University of Washington, Seattle; 2003.
13. Dervisoglu E, Isgoren S, Kasgari D, Demir H, Yilmaz A. Obesity control and low protein diet preserve or even improve renal functions in Bardet–Biedl syndrome: a report of two cases. Med Sci Monit. 2011;17:12–4. https:// doi.org/10.12659/MSM.881320.
14. Hooda AK, Karan SC, Bishnoi JS, Nandwani A, Sinha T. Renal transplant in a child with Bardet–Biedl syndrome: a rare cause of end-stage renal disease. Indian J Nephrol. 2009;19(3):112–4.
15. Mujahid S, Hunt KF, Cheah YS, Forsythe E, Hazlehurst JM, Sparks K, Mohammed S, Tomlinson JW, Amiel SA, Carroll PV, Beales PL, Huda MSB, McGowan BM. The endocrine and metabolic characteristics of a large Bardet–Biedl syndrome clinic population. J Clin Endocrinol Metab. 2018;103(5):1834–41. https://doi.org/10.1210/jc.2017-01459.

16. Kumar S, Mahajan BB, Mittal J. Bardet–Biedl syndrome: a rare case report from north India. Indian J Dermatol Venereol Leprol. 2012; 78(2): 22.