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Bardet–Biedl syndrome with steatohepatitis – A rare case

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Abstract

Bardet–Biedl syndrome is a rare, autosomal recessive multisystem disorder. An 11-year-old male presented to the ophthalmic outpatient department in a tertiary health center of Central India with complaints of diminution of vision in both eyes, especially at night, difficulty in speech, and abdominal distension. He also had a history of delayed developmental milestones. On general examination, he had truncal obesity, polydactyly, and hypogonadism. Abdominal ultrasound revealed cholelithiasis with nodular fatty liver. On dental examination, he had microglossia and high-arched palate. Pure-tone audiometry showed normal hearing sensitivity. Psychiatric evaluation revealed severe mental retardation. A comprehensive ophthalmic examination showed a visual acuity of 20/240 both eyes, telecanthus with prominent epicanthal folds in both eyes, depressed nasal bridge, and sparse eyebrows and eyelashes. Fundus examination showed retinitis pigmentosa. OCT revealed generalized retinal thinning and reduced foveolar depression. Laboratory examination including complete blood count, urinalysis, renal function tests, and thyroid function tests was within normal limits. Since there is no proven effective treatment to prevent or alleviate vision deterioration in these patients, spectacles and low vision aids were prescribed, and regular ophthalmological follow-up along with screening of family members was advised.

Keywords: Truncal obesity, Polydactyly, Retinitis pigmentosa, Hypogonadism, Hepatomegaly, Cholelithiasis

1. Introduction

Bardet–Biedl syndrome (BBS) is a rare autosomal recessive disorder characterized by cardinal symptoms of marked truncal obesity, retinal dystrophy, polydactyly, mental retardation, and hypogonadism.[1] Other less common features include hepatic fibrosis, diabetes mellitus, speech and language deficits, facial dysmorphism, dental anomalies, and developmental delay.[2] The frequency of BBS varies depending on geographic location and the level of consanguinity in the population, with higher prevalence in populations with high rates of consanguinity according to research studies. Less than 15 cases have been reported from India.[3] Male-to-female ratio is 1.3:1. The patients generally have onset of symptoms within the first 10 years of life with earliest complaint usually being of poor night vision. Peripheral visual fields are constricted and fundus shows constricted arterioles, waxy disc pallor with pigment atrophy, and bony spicule pigmentation in the periphery. Diagnosis is important for visual prognosis and low vision management.

We report here a typical case of BBS in an 11-year-old boy with additional findings of nonalcoholic steatohepatitis.

2. Case History

An 11-year-old male presented to the Ophthalmology outpatient department with presenting complaints of inability to see properly with frequent falls while walking, more frequently at night, difficulty in speech, abdominal distension, and limping on the left leg. He was born of a consanguineous marriage as a full-term normal vaginal delivery but with delayed cry. The mother had no history of any antenatal medications and the baby was completely immunized as per the vaccination schedule. The child had delayed physical and mental growth with delayed developmental milestones, as shown in Table 1.

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On physical examination, the child had moon facies, dull-looking appearance, central obesity, depressed nasal bridge, and postaxial polydactyly in both hands and both feet [Figure 1a and b]. His blood pressure was 110/70 mmHg, height and weight were 130 cm and 65 kg, respectively, with resultant body mass index of 38.4, indicating severe obesity [Figure 1a], and proximal lower limb weakness with power grade 3. He also had hypogonadism with micropenis (<2.5 cm) [Figure 1d], testicular volume of 2 mL, and absence of secondary sexual characteristics.

Dental examination revealed microglossia and high-arched palate [Figure 1c]. On ears, nose, throat evaluation, the patient was diagnosed with delayed speech and language development, but with bilateral normal hearing sensitivity by pure-tone audiometry. Psychiatric assessment showed severe mental retardation with an IQ of 24.

On ophthalmological evaluation, the child had visual acuity 20/240 in both eyes. He also had hypertelorism and prominent epicanthal folds in both eyes. Fundus examination showed a waxy pale disc with marked attenuation of arterioles and scanty bony spicular pigments in periphery, suggestive of early retinitis pigmentosa in both eyes [Figure 1e and f]. Visual field analysis by confrontation showed bilateral constriction of visual fields, and OCT in both eyes revealed generalized retinal thinning and reduced foveolar depression.

His abdominal ultrasound showed cholelithiasis with hepatomegaly and nodular fatty liver. The cardiovascular and respiratory system showed no significant abnormality with normal echocardiography (ECG), ECG, and X-ray chest. Magnetic resonance imaging brain revealed a normal study. Laboratory examinations including complete blood count, urinalysis, liver function tests, renal function tests, and thyroid function tests were found to be normal.

Table 1: Developmental milestones

Milestone	**Normal age**	**Patient age**
Neck holding	3 months	10 months
Rolls over	5 months	12 months
Unidextrous reach	6 months	16 months
Monosyllables	6 months	6 years
Sitting without support	8 months	2 years
Standing with support	9 months	30 months
Walking	15 months	3 years
Running	18 months	5 years

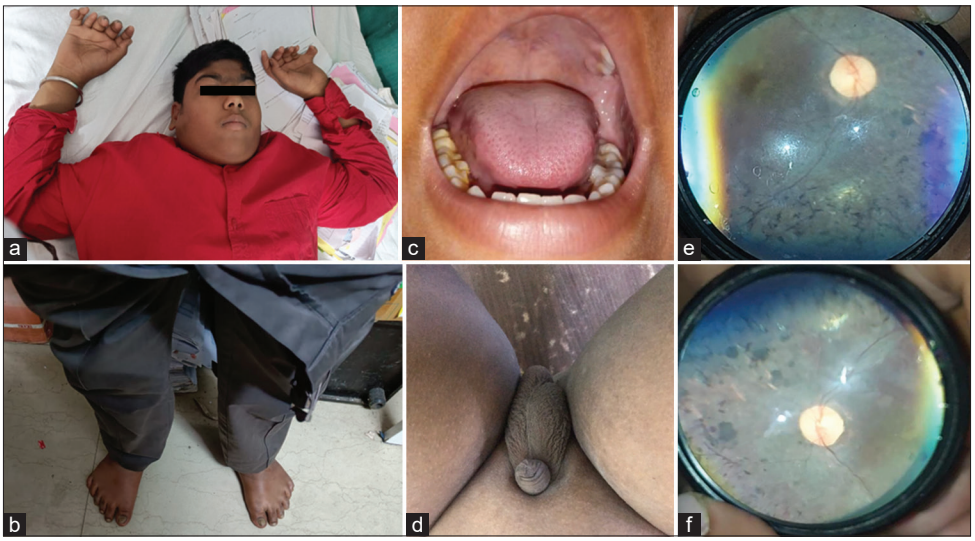

Figure 1: Clinical manifestations

3. Discussion

BBS is named after George Louis Bardet, a French physician, and Arthur Biedl, a Hungarian pathologist in 1920. It is an inherited autosomal recessive ciliopathic human genetic disorder.[4] Parents must be carriers of the defective gene and both must pass on the defective gene to child for the child to get affected. If both individuals carry the defective gene, which is responsible for causing BBS, there is a 25% chance of having a child with this syndrome, 50% chance to be an affected carrier, and a 25% chance to be unaffected and not a carrier.[5]

Around 19 genes are found responsible for Bardet–Biedl syndrome. Pathogenic variants in 14 genes are known to be associated with BBS. About quarter of all cases result from mutation of *BBS1* gene located on chromosome 11q14 and mutation in *BBS10* gene is responsible for another 20% cases. The remaining *BBS* genes are responsible for only very few cases and in about a quarter of cases, the cause of this syndrome is unknown. Gene products encoded by *BBS* genes called BBS proteins assist microtubule intraflagellar transport and cellular organization.[6] Mutations of *BBS* genes result in defective intra-flagellar transport.[7]

Diagnosis of BBS is established by clinical findings which can be grouped under primary features and secondary features. The main primary features are retinitis pigmentosa, postaxial polydactyly, truncal obesity, hypogonadism, learning difficulties, and renal abnormalities. Secondary features include speech and developmental delay, behavioral abnormalities, ataxia, poor coordination, hypertonia, diabetes mellitus, orodental abnormalities, cardiovascular anomalies, hepatic disease, craniofacial dysmorphism, Hirschsprung disease, and anosmia. Patients who have four primary features, or three primary features and two secondary features, are diagnosed with Bardet–Biedl syndrome.[8]

Our 11-year-old male patient with Bardet–Biedl syndrome had four primary features (retinitis pigmentosa, postaxial polydactyly, truncal obesity, and hypogonadism) and four secondary features (speech and developmental delay, high arched palate, dental abnormalities, and steatohepatitis).

BBS has an adverse prognosis and treatment depends on clinical features and organs involved. A multidisciplinary approach is needed for managing this syndrome based on clinical manifestations. Treatment may require coordinated efforts of team of ophthalmologists, pediatricians, dental specialists, speech pathologists, audiologists, and other healthcare professionals. Regular monitoring of renal, liver, glucose, lipid, and endocrine profile is necessary. Testosterone supplements may be prescribed to male patients in cases having lowered level of this hormone. Accessory digits are often non-functional and may be excised. A low-calorie and low-protein diet helps in obesity control. Educational programs to overcome learning disabilities are important. There are no proven effective treatments to either prevent or alleviate deterioration in vision. However, spectacles and low vision aids can be prescribed and regular ophthalmological follow-up should be advised. Psychological counseling is of benefit for affected individuals and their families.

In view of its autosomal recessive inheritance, genetic counseling, informed reproductive choices, and early screening of at-risk family members should be advised. Furthermore, ultrasound screening in the second trimester for polydactyly and renal anomalies in high-risk pregnancies can help in the antenatal diagnosis of Bardet–Biedl syndrome.

4. Conclusion

BBS is extremely rare disorder that affects multiple organs in human body. Diagnosis is mainly based on clinical features and can be confirmed by genetic testing. Early diagnosis and multidisciplinary approach in treatment is necessary to delay the complications and to enhance quality of life of patients. In view of its autosomal recessive inheritance, genetic counseling, informed reproductive choices and early screening of at-risk family members should be advised. Further research is needed to study the potential genetic factors contributing to complex presentation and to enhance our understanding of this rare genetic disorder.

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