

Bardet- Biedl syndrome – A rare case

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Abstract

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive disorder with wide spectrum of clinical features. BBS is distinguished from the much rarer Laurence-moon syndrome, in which retinal pigmentary degeneration, mental retardation, and hypogonadism occur in association with progressive spastic paraparesis and distal muscle weakness, but without polydactyly. Most common feature of BBS is retinal dystrophy. The visual prognosis for children with Bardet-Biedl syndrome is poor.

Keywords: Mental retardation, Pigmentary retinopathy, Polydactyly.

Introduction

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive disorder with wide spectrum of clinical features that produces many effects and affects many body systems. Its main features are obesity, pigmentary retinopathy, polydactyly, hypogonadism and mental retardation and renal dysfunction.¹ BBS expression varies both within and between families and diagnosis is often difficult. The average age at diagnosis is 9 years. Diagnosis is made by clinical findings. Twelve genes are known to be associated with BBS.²

Case Report

An 11 years old boy presented with chief complaint of decreased vision mainly in the night. There was no history of any consanguineous marriage between parents. The birth of the patient was normal. He had learning disability from birth.

On systemic examination patient was obese with BMI (30.3kg/m²), and mental development was lagged behind the normal range. There was presence of polydactyl (hexadactyly) (Fig. 1), hypogonadism but auditory and cardiovascular systems were within normal limits.

On ophthalmic examination patient was orthophoric, ocular movements were full in all directions of gaze in both the eyes.

The best correction visual acuity was counting finger 5 meters in both the eyes. On slit lamp examination of anterior segment of eye was within normal limits. Fundus examination showed bulls eye maculopathy with mid peripheral pigmentary changes with bony spicules, arteriolar constriction with mild pallor of the disc (waxy) (Fig. 2 and Fig. 3).

Laboratory investigation, urine examination, kidney function tests, complete hemogram with ESR were within normal limits. Chest and abdominal X-ray were normal. Abdominal USG and ECG were also within normal limits.

Discussion

Laurence-moon (Bardet) Biedl syndrome is a rare disorder first defined by Bardet in 1920.³ BBS is distinguished from the much rarer Laurence-moon syndrome, in which retinal pigmentary degeneration, mental retardation, and hypogonadism occur in association with progressive spastic paraparesis and distal muscle weakness, but without polydactyly.⁴



Fig. 1: Polydactyly of both the feet

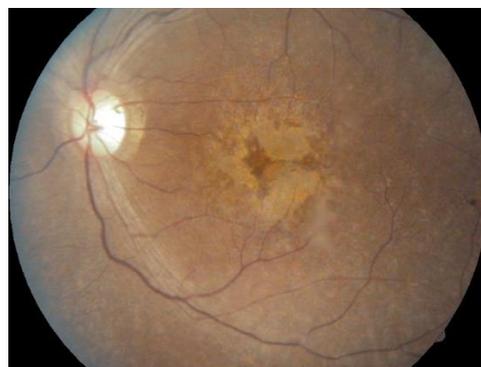


Fig. 2: Fundus of left eye showing bulls eye maculopathy

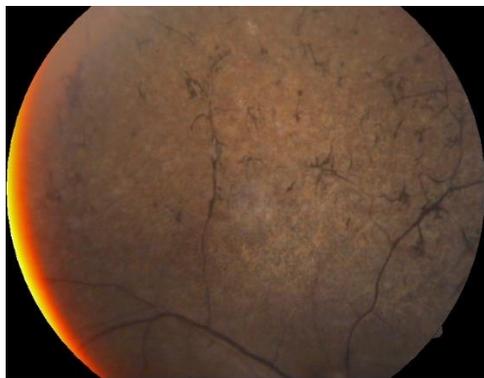


Fig. 3 Fundus showing bony spicules mainly in the mid periphery

Most common feature of BBS is retinal dystrophy. The retinal appearance is variable, with typical retinitis pigmentosa may be present in only few cases.⁵ Other features of BBS are obesity, polydactyly, mental retardation, hypogonadism and renal dysfunction. Other associated features can be hepatic fibrosis, diabetes mellitus, reproductive abnormalities, endocrinological abnormalities, short stature, developmental delay and speech deficit, not always present.

Patients generally has onset of symptoms within the first 10 years life and most often the first complaint is poor night vision.⁶ Nystgmus may be a common feature. The visual prognosis for children with Bardet-Biedl syndrome is poor, progressive loss of visual acuity arises early in life in the first decade of life.

Conclusion

The index case has many common features of Laurence Moon Bardet Biedl syndrome including polydactyly, obesity, mental retardation, hypogonadism and retinitis pigmentosa.

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