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Spot Diagnosis of a Daedalian Genetic Disorder: Bardet-Biedl Syndrome

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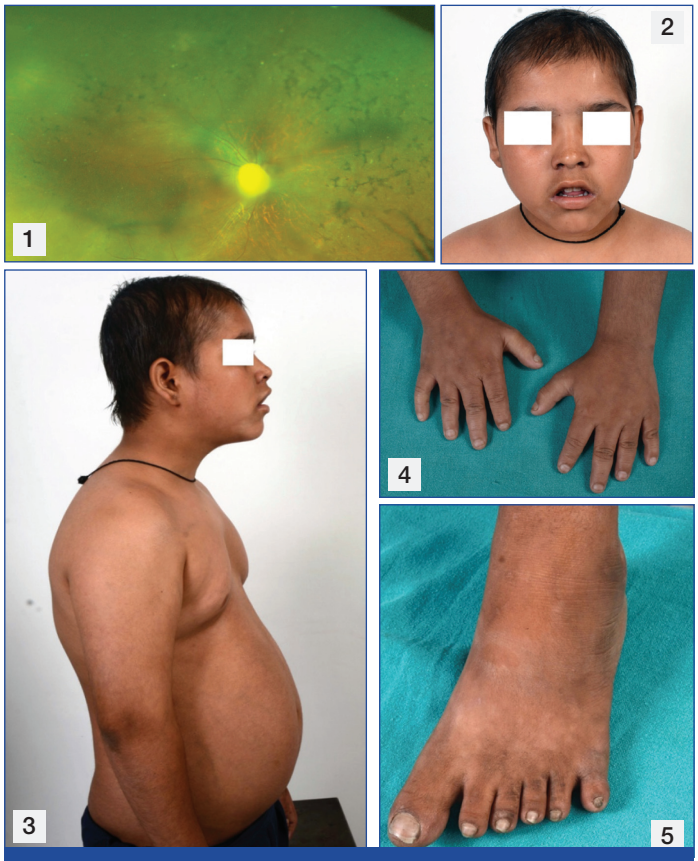
A 15-year-old boy with night blindness since the age of six presented to the outpatient wing with a history of growth retardation, poor scholastic performance and failure to attain secondary sexual characters. Retinal imaging showed the presence of pigmentary retinal degeneration and attenuated vessels suggesting retinitis pigmentosa [Table/Fig-1]. Physical examination revealed dysmorphic facies [Table/Fig-2] - widened intercanthal distance, low set ears, depressed nasal bridge and a flattened occiput [Table/Fig-3]. Also truncal obesity [Table/Fig-3], shortened metacarpals/phalanges [Table/Fig-4], polydactyly [Table/Fig-5], and underdeveloped testes were noted. In view of the constellation of findings, a diagnosis of Bardet–Biedl Syndrome (BBS) was made. Nutritional supplementation was advised and the patient was referred for IQ assessment and genetic counseling.

In 1866, Laurence and Moon described four patients with retinal degeneration, obesity, and cognitive deficit [1]. Bardet and Biedl separately reported similarly affected individuals who also had postaxial polydactyly and the condition was coined Laurence–Moon–Bardet–Biedl syndrome [2,3]. The syndrome is often divided into two entities: Laurence–Moon syndrome and Bardet–Biedl Syndrome (BBS), but since there is considerable overlap, BBS is now the globally accepted name.

Beales et al., proposed a modified criteria in 1999, requiring four primary features or three primary features with two secondary features for the diagnosis [4]. Rod-cone dystrophy, polydactyly, truncal obesity, genital anomalies, chronic kidney disease and learning disabilities form the primary features, while speech delay, developmental delay, diabetes, dental anomalies, congenital heart disease, brachydactyly/syndactyly, ataxia/poor coordination and anosmia/hyposmia are the secondary features. The diagnosis of BBS is delayed and considered only when the child develops visual problems associated with rod-cone dystrophy [5]. Postaxial polydactyly may be the only hint to the diagnosis at birth and hence its presence should alert physicians to look for other features of BBS [5]. Our patient had five primary and three secondary features.

BBS is a rare and complex syndrome, autosomal recessively inherited, involving mutations of proteins that form components of the ciliary apparatus. But as emphasized by our patient, the diagnosis remains clinical. Treatment and rehabilitation involve a multidisciplinary approach. Early recognition and treatment of co-morbidities are imperative to improve the quality of life.



[Table/Fig-1]: Fundus photograph of the left eye demonstrating pigmentary changes and arteriolar attenuation characteristic of retinitis pigmentosa.
[Table/Fig-2]: Dysmorphic face with depressed nasal bridge, widened intercanthal distance and low set ears. **[Table/Fig-3]:** Depicting the flattened occiput and trucal obesity. **[Table/Fig-4]:** Brachydactyly, shortened metacarpals and phalanges. **[Table/Fig-5]:** Postaxial polydactyly.

REFERENCES

[1] Laurence JZ, Moon RC. Four cases of "retinitis pigmentosa" occurring in the same family, and accompanied by general imperfections of development 1866. *Obes Res*. 1995;3:400-03.
[2] Bardet G. On congenital obesity syndrome with polydactyly and retinitis pigmentosa (a contribution to the study of clinical forms of hypophyseal obesity) 1920. *Obes Res*. 1995;3:387-99.
[3] Biedl A. A pair of siblings with adiposo-genital dystrophy 1922. *Obes Res*. 1995;3:404.
[4] Beales PL, Elcioglu N, Woolf AS, et al. New criteria for improved diagnosis of Bardet-Biedl syndrome: results of a population survey. *J Med Genet*. 1999;36:437-46.
[5] Forsythe E, Beales PL. Bardet-Biedl syndrome. *Eur J Hum Genet*. 2013;21:8-13.

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