International Journal of Contemporary Pediatrics
Tiwari K et al. Int J Contemp Pediatr. 2016 Nov;3(4):1480-1482
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20163707

Case Report

Bardet biedl syndrome: a rare occurrence

Kavita Tiwari*, Suresh Meena, Suresh Goyal

Department of Pediatrics, RNT Medical College, Udaipur, Rajasthan, India

Received: 03 September 2016
Accepted: 04 October 2016

***Correspondence:**
Dr. Kavita Tiwari,
E-mail: renaissance_28@yahoo.co.in

ABSTRACT

The bardet-biedl syndrome (BBS) is a rare autosomal recessive genetic disorder that affects many body systems. It is characterized principally by obesity, retinitis pigmentosa, polydactyly, hypogonadism, kidney abnormalities and learning difficulties. We hereby present a 14 year old male patient exhibiting characteristic features of bardet biedl syndrome (BBS) along with a brief review of the literature.

Keywords: BBS, Obesity, Polydactyly, Retinitis pigmentosa

INTRODUCTION

Bardet biedl syndrome (BBS) is a rare autosomal recessive disorder. BBS was first described by Bardet and Biedl in the 1920.[1] The principal manifestations are rod-cone dystrophy (retinitis pigmentosa), postaxial polydactyly, central obesity, mental retardation, hypogonadism, and renal dysfunction. Other features not always present include hepatic fibrosis, diabetes mellitus, neurological, speech and language deficits, behavioral traits, facial dysmorphism, dental anomalies and developmental delay.[2] The frequency of the syndrome is estimated to be 1:1,40,000 o 1:1,60,000.[3] So far, very few cases have been reported from India.

CASE REPORT

A 14 year old boy reported to outpatient department of Paediatrics with complaint of headache. Presence of obesity, striae, hypertension and bilateral blindness at first made us to thought about cushings disease. But on thorough examination patient was found to have truncal obesity (weight 72 kgs) with BMI of 30kg/m^2, underdeveloped genitalia, polydactyly, brachydactyly of both hands, with a history of complete painless loss of vision in both eyes from last 3 years along with learning difficulties with near normal gross motor, fine motor and social development. He was a product of non consanguinous marriage with 5 other siblings among which two died in infancy due to some acute illness. His BP was 140/94 mmHg.

Lab investigations revealed hypercholesterolemia, other investigations like hemogram, RFT, LFT and routine urine examination were normal. Fundus examination revealed arteriolar attenuation, pallor of the optic disc,bone-spicules and pigment clumping (findings of retinitis pigmentosa). MRI brain (done initially thinking of cushings disease) was normal. USG whole abdomen and ECHO were reported normal.

DISCUSSION

Bardet-biedl syndrome shows significant overlap with a disorder called Laurence-moon syndrome. In fact, in the past these disorders were considered the same and referred to as Laurence-bardet-biedl syndrome. BBS is distinguished from the much rarer Laurence- Moon syndrome, in which retinal pigmentary degeneration, mental retardation and hypogonadism occur in

conjunction with progressive spastic paraparesis and distal muscle weakness, but without polydactyly.[2]

Table 1: Modified diagnostic criteria and clinical manifestation present in our case.

Primary features	Case
Rod-cone dystrophy	+
Polydactyly	+
Obesity	+
Learning disabilities	+
Hypogonadism in males	+
Renal anomalies	-
Secondary features	
Speech disorder/delay	+
Strabismus/cataracts/astigmatism	+
Brachydactyly/ syndactyly	+
Developmental delay	+
Nephrogenic diabetes insipidus	-
Ataxia/poor coordination/imbalance	+
Mild spasticity	-
Diabetes mellitus	-
Dental crowding/hypodontia/small roots	-
Left ventricular hypertrophy/congenital heart disease	-
Hepatic fibrosis	-

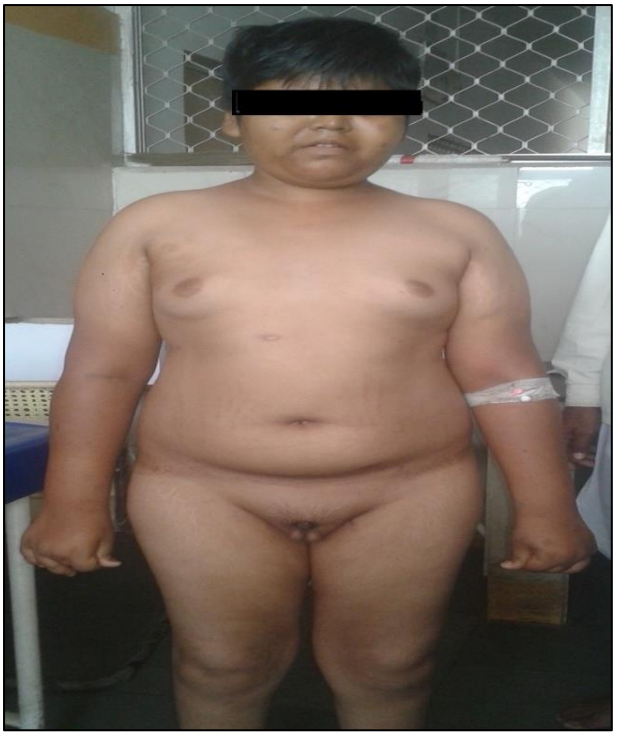

Figure 1: 14 year old male child with bardet beidl syndrome having obesity and hypogonadism.

Bardet-biedl syndrome is a rare, genetic multisystem disorder characterized primarily by deterioration of cone and rod cells in the retina of the eyes (progressive cone-rod dystrophy), an extra finger or an toe near the fifth toe (postaxial polydactyly), fat is disproportionately distributed on the abdomen and chest rather than the arms and legs (truncal obesity), diminished size and decreased function of the gonads (testes), in males (hypgonadism), renal abnormalities, and learning difficulties.

Visual abnormalities usually become progressively worse and may ultimately result in blindness. Kidney abnormalities may progress to cause life-threatening complications. Only a minority of affected individuals have severe mental impairment.[5] In 1999, modified diagnostic criteria were defined after a study conducted in England in 109 BBS patients.[4] Patients who had 4 primary characteristics or 3 primary and 2 secondary criteria were identified as BBS (Table 1).

Thus our case had 5 primary and 5 secondary features present characteristic of the syndrome. The diagnosis had been missed until the patient presented at our hospital.

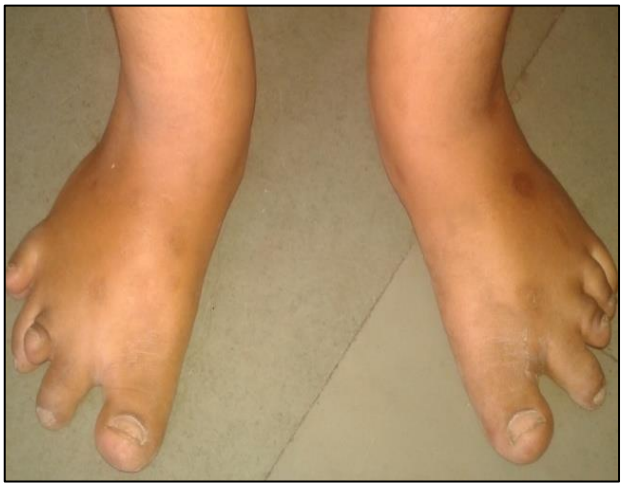

Figure 2: Feet of bardet biedl syndrome patient having syndactyly of third and fourth toe.

CONCLUSION

The multisystem involvement of this disorder exemplifies the need for multidisciplinary management in such cases. The case is being reported for its rarity as well as an early identification of the syndrome may help in prevention of and early treatment of hypertention, blindness and other comorbidities which can increase the productive lifespan in such cases.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Jones KL. Miscellaneous syndromes: bardet biedl syndrome. Smith's Recogniable Patterns of Human Malformation. Sixth Edition: Elsevier Health. 2013:676-677.

2. Haque MA, Sharmin LS, Islam QT, Ekram ARMS. Bardet biedl syndrome- a case report. Journal Teachers Asso. 2007;20:56-9.
3. Vanikar AV, Gumber MR, Patel HV, Shah PR, Patil SB, Trivedi HL. Bardet-biedl syndrome: a rare cause of chronic kidney disease. Indian J Clin Biochem. 2013;28:201-5.
4. 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;36:437-46.
5. Kumar S, Mahajan BB, Mittal J. Bardet-biedl syndrome: a rare case report from North India. Search Results. Indian J Dermatol Venereol Leprol. 2012;78:228.

Cite this article as: Tiwari K, Meena S, Goyal S. Bardet biedl syndrome: a rare occurrence. Int J Contemp Pediatr 2016;3:1480-2.