Case Report

Bardet-Biedl Syndrome – A Case Report

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ABSTRACT

An 8 year old girl was brought to the ophthalmology OPD with complaints of reduced vision and night blindness. She was born with polydactyly in the both feet and one hand. She had developmental delay, mental retardation, learning difficulty. On examination she had polydactyly, moon face, obesity. Retinitis pigmentosa was found on fundoscopy. With typical features, she was diagnosed as a case of Bardet-Biedl syndrome. It is commonly found in communities with high inter-family marriage. Clinical features appear early in childhood and diagnosis is usually done by puberty. Prominent features include rod-cone dystrophy leading to blindness, postaxial polydactyly, central obesity, learning disability, hypogonadism in males and renal dysfunction. Relatives with a single affected gene may have obesity, hypertension, diabetes and renal disease. There is increased risk of renal involvement. There is no definite treatment. Early diagnosis and symptomatic, supportive and rehabilitative measures can reduce the disability.

Keywords: Bardet-Biedl syndrome, polydactyly, retinitis pigmentosa, hypogonadism

INTRODUCTION

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive disorder with clinical and genetic heterogeneity. The main features are obesity, polydactyly, pigmentary retinopathy, learning disabilities, various degrees of intellectual impairment, hypogonadism and renal abnormalities. The diagnosis of Bardet-Biedl syndrome (BBS) is established by the clinical findings.

CASE REPORT

An 8 year old girl was brought to the ophthalmology OPD with complaints of reduced vision and night blindness. On examination she was not cooperative for visual acuity recording but mother told that she can count fingers from 2 metres distance. She had mild exotropia. Anterior segment appeared normal in both eyes. Fundus examination showed features of atypical retinitis pigmentosa(fig-1).

History of parental consanguineous marriage is present and no relatives known to have the disease. The birth of the patient had been normal. She had delayed developmental mile stones and learning difficulties. Physical development was within the normal range of chronological age, but mental development was lagged behind the normal range.

On general examination patient had frontal bossing, flat nasal bridge, short neck, obesity(fig-2), polydactyly in right hand and both feet(fig-3). The vital signs were within normal limits.Physical examination showed height 122 cm, weight 35kg.

Routine blood investigations, USG abdomen and 2D echo were done and all are found to be with in normal limits. Visual fields could not be taken as patient is not co-operative.

With typical features, she was diagnosed as a case of Bardet-Biedl syndrome.

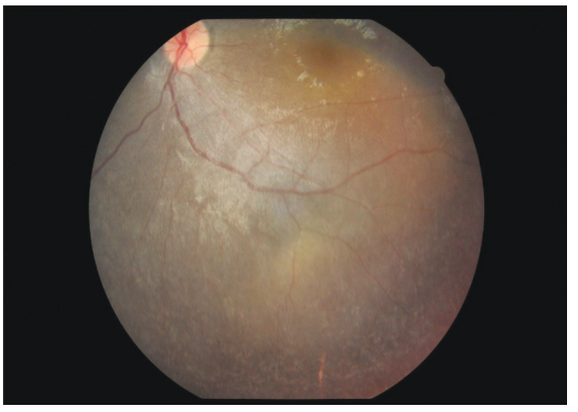

Figure 1 : Fundus picture showing atypical retinitis pigmentosa

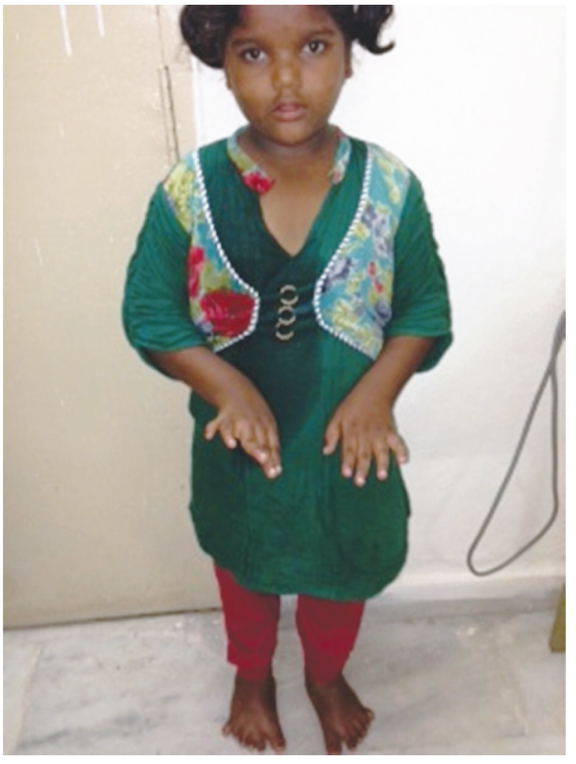

Figure 2: Central obesity

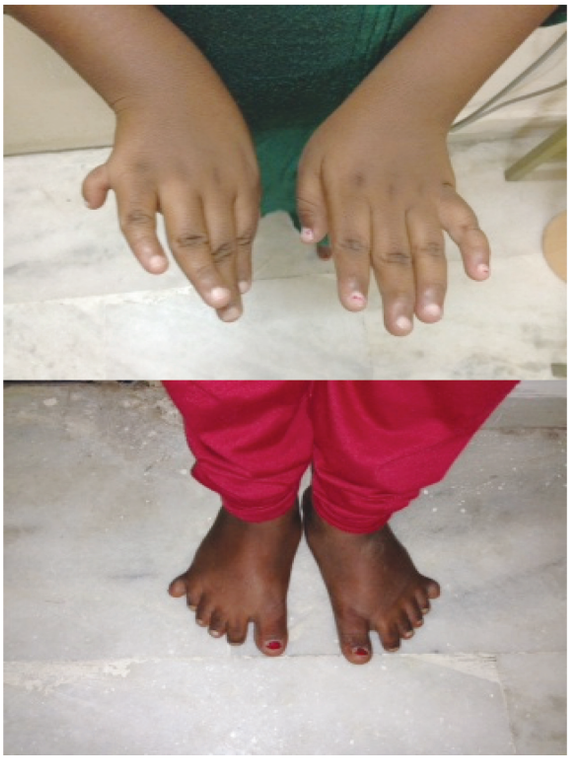

Figure 3: Polydactyly in one hand and both feet

DISCUSSION

The variable manifestations of Bardet-Biedl syndrome (BBS) were initially described by Bardet and Biedl in the 1920s [1]. It is clearly different from a condition reported by Lourence and Moon in 1865. If spasticity and mental retardation are present, it fulfills the criteria for Laurence-Moon-Bardet-Biedlsyndrome[2]. The BBS phenotype is seen in individuals with mutations in 11 different genes [3, 4]. BBS1 accounts for 25-30% of cases. The exact pathogenesis of BBS is unknown. It has been recently recognized that proteins coded for by the BBS4, BBS6, BBS8, and BBS10 genes are expressed in the basal body of cilia,[3, 5, 6, 7] and BBS is now regarded as one of the 'ciliopathies.' The gene products are probably involved in the signaling pathway in the cilia; abnormalities interfere with normal development, resulting in the diverse pathological effects of the syndrome.[8, 9]

The cardinal manifestations of BBS are limb abnormalities, most commonly postaxial polydactyly (70-90%), pigmentary retinopathy (90-100%), central obesity (70-95%), and renal involvement (25-100%).[10] Less common features include male hypogonadism, mental retardation, behavioral abnormalities, hearing loss, speech disturbances, hypertension, type 2 diabetes mellitus, congenital heart disease, and cardiomyopathy. Hypogonadism, which is probably primary in origin, is reported more frequently in males than in females. [11] Rare associations include hypothyroidism, Hirschsprung's disease, epilepsy, genital anomalies, anal stenosis, and abnormal dentition. On the basis of a review of 109 cases, Bealeset al .[12] suggested the diagnostic criteria summarized in [Table - 1]

Table 1: Diagnostic criteria* for Bardet-Biedl syndrome

Primary features: Retinitis pigmentosa Postaxial polydactyly Central obesity Learning disabilities Renal disease

Secondary features:

Speech disorder / delay developmental
Strabismus / cataract / astigmatism
Brachydactyly / syndactyly
Developmental delay
Polyuria / polydipsia (nephrogenic diabeties insipidus)
Ataxia / poor coordination / imbalance
Mild spasticity (especially in lower limbs)
Diabetes mellitus
Dental crowding / hypodontia / small roots / high-arched palate
Left ventricular hypertrophy / congenital heart disease
Hepatic fibrosis

"Four primary, or three primary and two secondary features, are required for diagnosis

CONCLUSION

There is no definite treatment. Early diagnosis and symptomatic, supportive and rehabilitative measures can reduce the disability. Renal involvement is common and renal failure is most common cause of death in BBS. Close follow-up for renal involvement in patients with BBS and ALMS from an early age is highly recommended to prevent ESRD. The case is being reported for its rarity.

Conflict of Interest : None

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