Laurence-Moon-Bardet-Biedl Syndrome: A Case Report

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ABSTRACT

Laurence Moon Bardet Biedl syndrome is a rare autosomal recessive disorder, mostly seen in children born from consanguineous marriages. It is characterized by early onset retinal dystrophy, obesity, limb abnormality, mental retardation, hypogonadism and renal disease. In this report, we present a case of a 4-year-old boy who presented with complaints of night blindness. He had polydactyly, central obesity, hypogonadism, microdontia and retinal dystrophy. The patient was diagnosed with LMBBS.

Keywords: Laurence Moon Bardet Biedl syndrome (LMBBS), Retinal dystrophy, polydactyly

INTRODUCTION

Laurence-Moon-Bardet-Biedl Syndrome is an infrequent, genetically diversified, autosomal recessive disorganization presented early with clinical features like early onset night blindness with typical or atypical retinitis pigmentosa, post axial polydactyly, central obesity, mental retardation, hypogonadism and structural renal deformity.[1] Other minor defects include speech hamper or slow down, sensorineural hearing loss, ataxia, congenital heart problems as well as hepatic fibrosis. We report a case with typical phenotype and presence of similar complaints in a sibling.

CASE HISTORY AND EXAMINATION

A 4-year-old male child, product of non-consanguineous marriage, presented to the ophthalmology outpatient department of Sagarmatha Choudhary Eye hospital, Lahan, with chief complaints of decreased night vision. Parents gave history of similar complaints in the elder sister of the patient but she was not brought for examination.

Antenatal, natal and postnatal periods were uneventful. Developmental milestones were achieved except for decreased intelligence. Physical examination revealed presence of polydactyly and brachydactyly of both hands and feet. (Photograph A and D). There was central obesity (Photograph B), dental anomalies (Image C) features of hypogonadism and micro penis.

On ophthalmological examination, visual acuity was 6/60 in both eyes. Fundus examination revealed pallor of optic disc, bilateral attenuation of vessels and retinal pigmentary changes.

Image (Photograph)- A
DISCUSSION

In 1866, Laurence and Moon explained a case of a 7-year-old female with rod-cone dystrophy, hypogonadism, mental retardation, obesity and polydactyly. In 1920, Bardet described a 4-year-old female patient presented with rod-cone dystrophy, obesity, polydactyly and mental retardation. Two years after Bardet’s report, Biedl highlighted the complete scenario of clinical signs which includes skull abnormalities, anal atresia, mental deficiency and gastrointestinal disorders. Since these discoveries’ presence of above manifestations as union, often in children with normal mother and father (consanguineous marriage) has been called as Laurence-Moon-Bardet-Biedl syndrome (LMBBS).

The detailed biochemical process that causes BBS is still ambiguous. Twelve genes (BBS1 to BBS12) that are responsible for the disease have been identified. BBS proteins are components of centrosomes and affect the ciliary transport, hence the disease falls under the spectrum of ciliopathies.

Retinal dystrophy is most common major manifestation present in BBS. Patients usually present with night blindness which gradually progresses to blindness. Usually, night blindness is noticed at the mean age of 8.5 years but in our case, it was noticed earlier at the age of 4 years. Obesity usually begins in childhood and progresses with age. Limb abnormalities like post axial polydactyly, brachydactyly, of both hands and feet are common. Other major features include hypogonadism, mental retardation and renal dysfunction.

Multiple minor features have also been found and documented. Which consist delayed developmental milestones with growth, speech as well as language shortfall, psychosis, facial bones abnormality in developmental form, multiple pigmented nevi, hearing impaired, diabetes mellitus, cardiovascular abnormality, dental growth delay with dental abnormality, gastrointestinal stenosis or atresia and Hirschsprung’s disease.

LMBBS has an unfriendly prediction, with early age loss of vision (mostly night blindness), central obesity, hypertension and diabetes mellitus. LMBBS with visual impairment need low vision aids. Surveillance includes regular ophthalmological examination, blood pressure measurement, monitoring of renal function, and regular testing of lipid profile and diabetes mellitus.
CONCLUSION
Diagnosis of Bardet Biedl syndrome is mainly clinical as genotyping is not available in many places especially in developing countries like India and Nepal.
Multidisciplinary approach is required for management, to provide good prognosis and quality of life to our patients.

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