

Laurence Moon Bardet Biedl Syndrome: A Rare Case Report in a Tertiary Care Teaching Hospital, Aurangabad, Maharashtra

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Abstract: *Laurence Moon Bardet Biedl Syndrome is a rare Ciliopathic and Pleiotrophic human Autosomal recessive genetic disorder, which involves affects and effects on multiple organ system. Consanguineous marriage is usually the common cause. The characteristic feature of the disorder are progressive rod cone dystrophy, atypical retinitis Pigmentosa, myopia, central obesity, mental retardation, Anisometropia, Astigmatism, Postaxial Polydactyly, Hypogonadism in males, renal involvement. It affects males and females equally. The treatment of Laurence Moon Bardet Biedl Syndrome is usually directed towards the specific symptoms that are apparent in each individual. We here present a case report of 30 year old male patient presenting in medicine department with uncontrolled blood sugar levels, morbid obesity, progressive loss of vision and polydactyly*

Keywords: Obesity, Hypogonadism, Retinitis Pigmentosa, Para paresis, Mental retardation, rod cone dystrophy

1. Introduction

Laurence-Moon syndrome (LNMS) is a genetic condition that results in a complex association of problems that affect several different body parts. People with LNMS may have difficulties with functions of the brain, eyes, ears, stomach, kidneys, hands and feet. They often also demonstrate a tendency to short stature and obesity. LNMS was later termed Laurence-Moon-Bardet-Biedl syndrome because of similarities with Bardet-Biedl syndrome (BBS). These two disorders also share similarity to Oliver-McFarlane syndrome (OMS). All three conditions are characterized by progressive blindness, obesity, and learning disabilities.

LNMS is associated with difficulty in controlling body movements, which is also seen in adolescent and adult patients with OMS and less frequently in BBS. LNMS and OMS are associated with abnormal pituitary gland function. BBS is associated with extra fingers and toes (postaxial polydactyly), and kidney and liver dysfunction. In patients with LNMS, fingers and toes may however have some degree of webbing, which is referred to as "syndactyly" Less often, they may have brachydactyly. The feet may also be overall short in length, of wide width, and carry a flat arch. Patients with LNMS may experience taurodontism, in which the development of tooth's body is enlarged relative to the roots.

People with LNMS have problems with the coordination of their body's movements and present with ataxia and spasticity due to cerebellar dysfunction.

Most patients with LNMS will experience a gradual loss of vision. The term "retinitis pigmentosa" is used to describe the particular, gradual-onset, vision loss that progresses according to a particular pattern. Retinitis pigmentosa begins with a night blindness that worsens with a loss of the ability to distinguish colors from one another, finally deteriorating into "tunnel vision".

Mild-to-moderate learning difficulties are common in individuals with LNMS. If the learning disability is rooted in neurological impairments, they are often associated with symptoms of poor coordination, gross and fine motor skills, and social milestones in childhood such as inability to play complicated games with other children.

Hypogonadism and pituitary abnormalities are also common. Other less common features that have been reported in patients living with LNMS include a "brachycephaly" and electrical abnormalities of the heart. People may also experience a loss of hearing, increased incidence of diabetes, liver fibrosis, and urinary and genital structural malformations.

2. Case report

A 30 years old male patient presented with acute gastroenteritis in our department. Patient was a known case of diabetes mellitus since 4 years and hypertension on medication. Patient was morbidly obese on presentation with polydactyly and hypodactyly. These findings intrigued us to search for any other cause of his obesity. On enquiry patient also complained of diminished vision after 6 pm since childhood. The birth history of the patient revealed that he was born out of consanguineous marriage. His mother was normal during pregnancy and had no history of taking any medications during gestation period. He was born at full term by normal vaginal delivery. History of birth asphyxia, cyanosis, feeding difficulties was negative. The baby was immunized periodically. According to the patient's mother baby had delayed physical and mental growth with delayed developmental milestones. He started walking at the age of 4 years and spoke when he was around 5 years old. He gained weight with little or no delay. Genetic tests were not done because of the non-availability of the test in our institute and poor socioeconomic status of the patient.

Further physical examination revealed dental crowding, hypodontia, micropenis, learning disability.

BMI- 33.2 kg/sq.m

Other Laboratory investigation

Kidney functions

Serum Creatinine- 3.5 mg,
urea – 96 mg/dL,
sodium-144 meq/lit,
potassium -5.5 meq/lit

CBC

haemoglobin – 5.6 gm%,
total leucocyte count -1700,
platelet- 3.5lakhs

Liver Function

Serum bilirubin-0.9 mg/dL,
SGOT-41 IU/L,
Serum albumin-1.8 gm/dL

Lipid Profile

Sr. Cholesterol- 298 mg/dL
HDL- 40 mg /dL
LDL- 170 mg/dL
Triglyceride- 180 mg/dL

USG (abdomen + pelvis)-

liver- span -18.5 cm, hepatomegaly with grade 1 fatty liver
spleen – 13.4 cm, splenomegaly
right kidney -12.4 x 5.2 cm
left kidney - 12 x 5.4 cm
bilateral hydronephrosis and hydroureter left>right

Fundus examination –

disc –normal size and shape, waxy pale,
e/o multiple bony spicules scattered over all quadrants
Features s/o Retinitis Pigmentosa

Modified diagnostic criteria for Bardet-Biedl syndrome

Primary Features

Rod cone dystrophy
Polydactyly
Obesity
Learning disabilities
Hypogonadism in males

Secondary features

Speech disorder/delay
Strabismus/cataracts/ Astigmatism
Brachydactyly/Syndactyly
Developmental delay
Polyuria/Polydipsia(Nephrogenic Diabetes Insipidus)
Ataxia/poor coordination/imbalance
Mild spasticity
Diabetes mellitus
Dental crowding/Hypodontia/Small roots/High arched palate
Left ventricular hypertrophy/Congenital Heart disease
Hepatic fibrosis

3. Discussion

BBS is named after Georges Bardet and Arthur Biedl. The first known case was reported by Laurence and Moon in 1866. Laurence-Moon-Biedl-Bardet syndrome (LMBBS) is no longer considered as a valid term as patients of Laurence

and Moon had paraplegia but no polydactyly and obesity, which are the key elements of the BBS. Hence, Laurence-Moon syndrome is usually considered a separate entity. However, some recent research suggests that the two conditions may not be distinct.^[4]

Apart from the cardinal manifestations, other features of the BBS include various degrees of intellectual impairment, congenital heart block, brachycephaly, deafness and dental anomalies. The full spectrum of clinical features is found in only 40-45% of LMBBS cases.^[5] Hypogonadism, which is probably primary in origin, is reported more frequently in males than in females.^[6]

The detailed biochemical mechanism that leads to BBS is still unclear. Twelve genes (BBS1 to BBS12) that are responsible for the disease have been cloned. The BBS proteins are components of the centrosome and affect the ciliary transport; hence, the disease falls under the spectrum of "ciliopathies."^[7]

Puberty is a particularly stressful time for those with BBS, and referring the patient to an experienced counselor is especially helpful. testosterone supplements may be prescribed to male patients, specifically in cases having lowered level of this hormone. There are no proven effective treatments to either prevent or alleviate the deterioration in vision. However, spectacles were advised as low vision was present in our patient, and regular ophthalmological follow-up was stressed upon. Accessory digits are often nonfunctional and may be excised. Obesity is an area of major concern in these patients and, if left uncontrolled, will lead to multiple health problems. A low-calorie and low-protein diet help in obesity control and may slow the progression of renal failure in patients with BBS.^[8] The patient was advised diet control as per the dietician's recommendations, and physical exercises.

As structural renal changes were identified in our patient, he was advised half-yearly urinalysis, blood pressure and urea and creatinine levels. Any derangement in these renal function tests would warrant prompt referral to a nephrologist as end-stage renal disease is an important cause of morbidity

In general, BBS patients are happy and friendly children, and the society should not have a discriminatory attitude toward these children so that their quality of life may be improved.

4. Conclusion

Our patient presented with the classical phenotype of BBS. Even though the patient had been seen by several specialists in different facilities, the diagnosis had been missed, possibly because of the rarity of the condition. It was the hypogonadism and the absence of secondary sexual characters that brought the patient to our department, where the diagnosis was made. The case is being reported for its rarity.

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