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# Laurence Moon Bardet Biedl Syndrome with Atypical Syndromic Retinitis Pigmentosa-A Rare Case Report

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**Abstract** **Case Report**

Laurence Moon Bardet Biedl syndrome is a rare autosomal recessive condition listed in the NORD (National Organisation for Rare Disorders) with only less than 15 cases reported all over India, affects children born to consanguineous parents. Early onset retinal dystrophy, obesity, limb abnormalities, mental retardation, hypogonadism, and renal illness being a few symptoms. Here is a case of 19-year-old male with this rare genetic disorder who presented with complaints of night blindness and defective vision since 2 years. On ocular examination both eye fundus showed atypical retinitis pigmentosa. On systemic examination patient had central obesity, polydactyly, mental retardation, hypogonadism, speech disorder, developmental delay and ataxia.

**Keywords:** Laurence Moon Bardet Biedl syndrome (LMBBS), hypogonadism, polydactyly.



## INTRODUCTION

Laurence-Moon-Bardet-Biedl syndrome (LMBBS) is a rare ciliopathic, pleiotropic autosomal recessive disorder that affects children born from consanguineous marriages. It was first described by Laurence-Moon in 1886 and Bardet and Biedl described additional cases between 1920-1922.The initial signs of these patients appear in the first ten years of life, with impaired night vision being the most common. The clinical grounds for making a diagnosis include the existence of four primary features on their own or three primary features plus two supplementary features. Polydactyly, cone-rod dystrophy, Obesity, mental retardation, and hypogonadism, renal structural malformation, Speech impairment, Sensorineural hearing loss, Congenital cardiac issues, ataxia, Hepatic fibrosis are all symptoms of LMBBS.

## CASE REPORT

18year old male patient was brought to OPD by his mother with complaints of Night blindness since 2years which was painless and gradual, weakness of upper and lower limbs since 1week with no history of a fall or trauma. He also found it difficult to mix and take the food to the mouth since 2 days which is an evidence of a cerebellar defect. His mother claimed that he had been mentally retarded since childhood, citing his delayed physical and mental development. She stated that he attained all his milestones delayed. Furthermore, the infant was the result of a consanguineous union. He was born via normal vaginal delivery with no previous history of birth asphyxia, feeding difficulties, or cyanosis, and he was regularly immunised. Birth History revealed Full Term Normal Vaginal Delivery with no previous history of birth asphyxia, feeding difficulties, or cyanosis, and was regularly immunised. Patient had no significant past history.

General physical examination showed a young man with below average height for age oriented time, place and person with Blood pressure of 128/80mm of Hg and Pulse rate of78bpm.Patient also had other significant features which include-Central Obesity with B.M.I. -31 (WHO Class I Obesity), Speech abnormality and Developmental delay since birth, Mental Retardation with I.Q. of 50, Polydactyly of lower and upper limb, Hypogonadism (Tanner Staging-II) with absence of pubic and axillary hair. Patient also had Upper and lower limb weakness (MRC power grading 3/5) and Plantar reflex was Flexor. He also found it difficult to mix and take the food to the mouth in the past 2 days which is evidence of a cerebellar defect. On systemic examination, Cardiovascular system showed no abnormality, S1and S2 were normal, no murmurs were noted. Respiratory system examination elicited

normal vesicular breath sounds. Abdomen was soft and no organomegaly was noted. Moderate Mental retardation of I.Q.- 50 was noted, No neurological deficits noted.

On ocular examination right eye visual acuity was 6/24 improving with pinhole to 6/18 and left eye visual acuity was 6/36 improving with pinhole to 6/18.Head posture and Facial symmetry was normal. Primary gaze position was orthophoric in both eyes. Anterior Segment evaluation both eyes showed Bitot spot at temporal bulbar conjunctiva with clear cornea and normally shaped reactive pupils with clear lens. Fundus examination of both eyes revealed atypical retinitis pigmentosa with pale disc, arteriolar attenuation and whitish raised lesion present all over fundus more around macula.

Following a thorough general physical and slit lamp examination, an Optical Coherence Tomography scan was taken which revealed Cone-Rod dystrophy. The patient also underwent a few other systemic investigations with normal findings

Although there is no cure for this disorder, Laurence Moon Bardet Biedl Syndrome requires a multi-disciplinary, symptomatic treatment with early detection, supportive, and rehabilitative interventions which help to lessen disability. Patient was given T. Vitamin- A 2,00,000 IU in view of bitot spot and to slow down the progression of retinitis pigmentosa. Glasses for constant use in view of Defective Vision was also given. Patient was also advised Physiotherapy for muscle weakness and Psychiatric Counselling intending Behavioural Therapy. The patient's mother was instructed to rigorously adhere to the specified dietary modification plan considering his central obesity.

**Figure 1: Patient with Central Obesity**

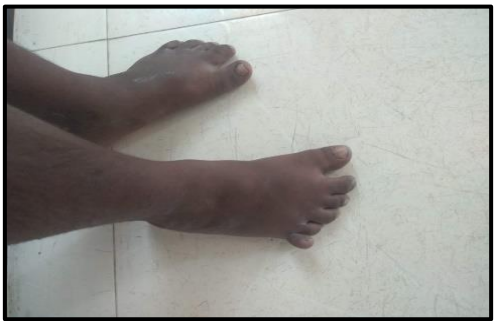

**Figure 2: Polydactyly of Right Foot**

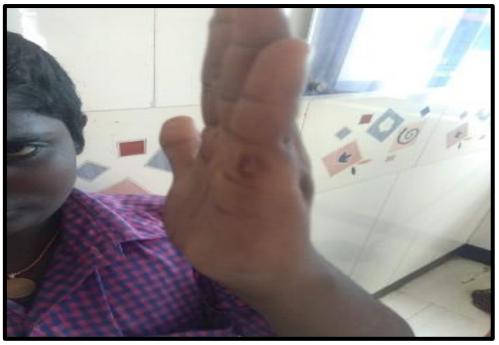

**Figure 3: Polydactyly-Nubbin of left hand**

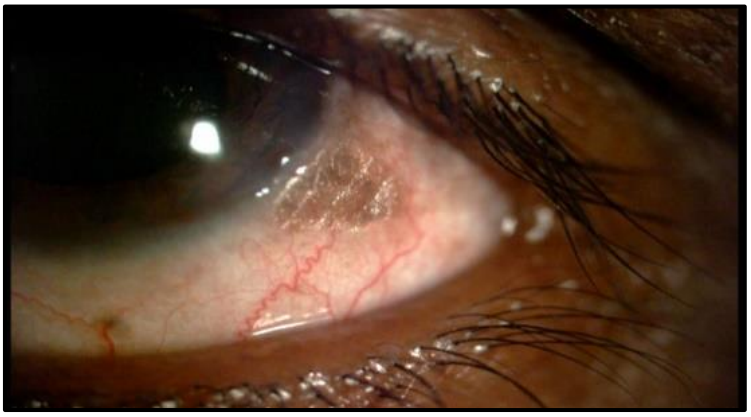

**Figure 4: Bitot Spot in Left Conjunctiva**

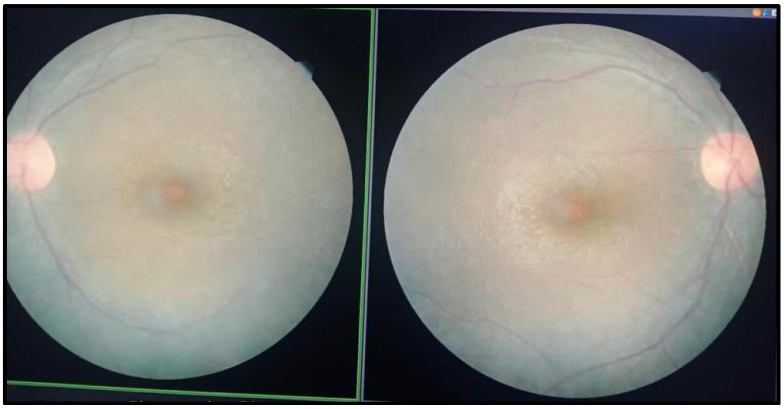

**Fig 5: Right and Left Eye Fundus picture**

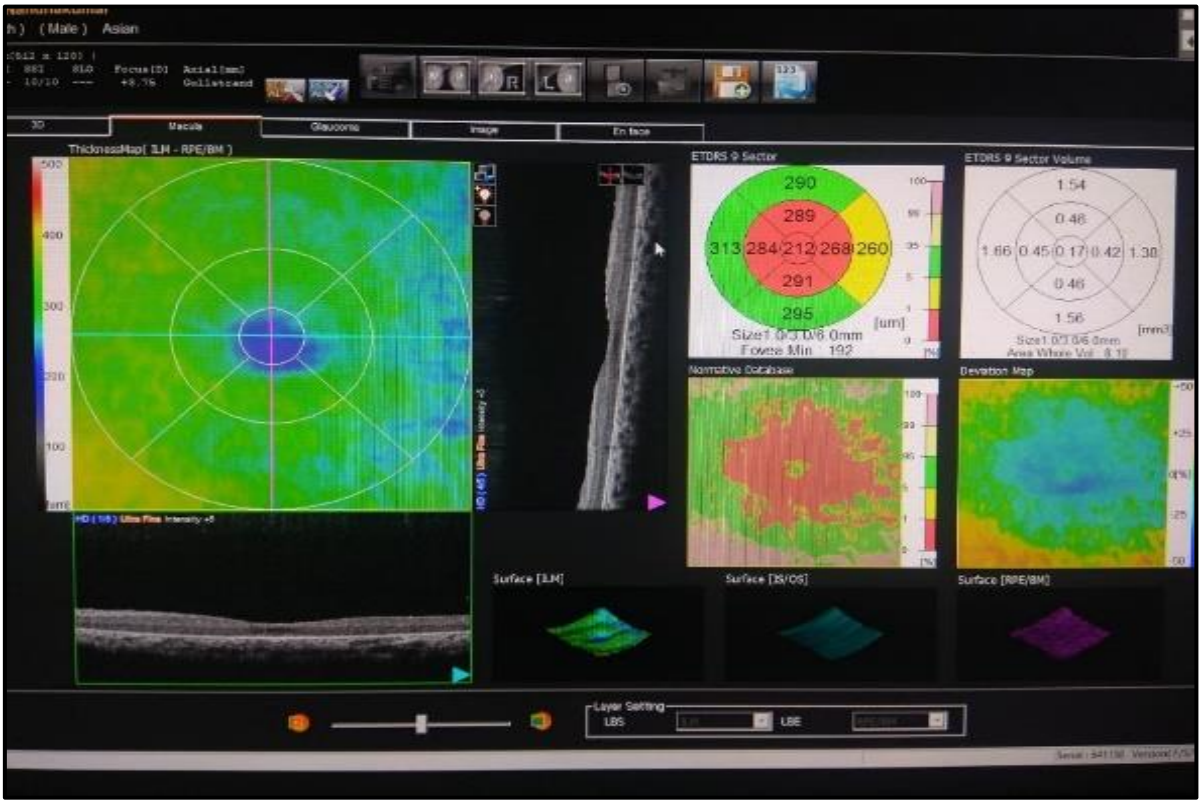


**Figure 6: OCT image showing Cone-Rod Dystrophy**

**Table 1: Systemic investigations**

| TEST | RESULT |
|---|---|
| Liver Function Test | Within Normal Limit |
| Complete Haemogram | Within Normal Limit |
| Renal Function Test and Urine Microscopy | Within Normal Limit |
| MRI Brain | Normal Study |
| USG Abdomen and Pelvis | No Abnormal Findings |
| ECG | Normal Study |
| ECHO | Normal Study |

## DISCUSSION

The reason for this study is the rarity of this syndrome with less than 15 cases reported all over India and hence the scarcity of information about LMBBS in India. The fact that our patient is a man, when the majority of documented cases are females also adds to the fact.

Laurence-Moon syndrome (LMS) and Bardet-Biedl syndrome (BBS) are still misunderstood amongst their presentation. Both have pigmentary retinal degeneration, mental retardation, and hypogonadism,

although LMS has more spastic paraplegia and BBS has more polydactyly and obesity. Some still consider BBS is a part of LMS because of some shared characteristics. BBS is a rare ciliopathic condition that affects 12 genes (BBS1 to BBS12), which produce proteins required for cilia function thereby creating structural and functional anomalies

**Table 2: Modified diagnostic criteria for Bardet-Biedl syndrome**

| Primary Features | Secondary Features |
|---|---|
| Rod-cone dystrophy, Polydactyly, Obesity, Learning disabilities, Hypogonadism in males, Renal anomalies. | 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. |

The treatments for LMBBS are mostly aimed at treating the symptoms of the disease. Physical treatment targeted at increasing strength is beneficial and exerciseas there is an increased incidence of diabetes and high cholesterol levels in individuals with LMS, a focused programme of nutritious, well-balanced diets and regular exercise is recommended. In BBS, a low-protein diet also decreases the progression of renal illness. Hormone replacement therapy can be used to treat the anterior pituitary gland's poor functional capability, which causes slow metabolism, poor growth, and infertility. Levothyroxine can help in body metabolism, hair-loss and tiredness. Growth hormone supplementation lessens the psychosocial impact of short height, testosterone supplementation helps underdeveloped genitalia in people with markedly low levels. Accessory digits are usually cosmetically unattractive and can be removed. Retinal dystrophy is usually the first symptom to appear before the age of ten years, but it affects nearly all patients under the age of twenty years. Glasses can help and seeing an ophthalmologist on a regular basis is advised.

Despite displaying LMBBS symptoms from a young age, our patient was not diagnosed until he was 16 years old. Doctors should refer such patients to a competent counsellor during puberty, as it is a tremendously difficult period for them, which our patient sadly missed.

## CONCLUSION

LMBBS causes significant morbidity and mortality in children. An early diagnosis can aid professionals in recognising and managing this problem more successfully. Because of its rarity and the involvement of several systems, it is frequently overlooked, even by experts. These youngsters would be able to better integrate into society and prosper if they were managed in a timely and thorough manner. Furthermore, either parents should get genetic counselling, particularly if the family has a history of consanguineous marriages. Outside-the-family marriages should likewise be encouraged. LMBBS patients are friendly children and the society should not have a discriminatory attitude toward these innocent children and if referred and diagnosed early their quality of life can certainly be improved.

## REFERENCES


- Genetic and Rare Diseases Information Center. Last updated: 6/18/2015 https://rarediseases.info.nih.gov/diseases/12635/laurence-moon-syndrome. Accessed May 25, 2018.
- Traboulsi, E. I. (2005). Compendium of Inherited Disorders and the Eye. New York: Oxford University Press.
- Farag, T. I., & Teebi, A. S. (1989). High incidence of Bardet Biedl syndrome among the Bedouin. *Clinical genetics*, *36*(6), 463-464.
- Emmanuel, M., & Bokor, B. R. (2021). Tanner Stages. [Updated 2020 Dec 18]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing.
- Beales, P. L., Elcioglu, N., Woolf, A. S., Parker, D., & Flinter, F. (1999). New criteria for improved diagnosis of Bardet-Biedl syndrome: results of a population survey. *Journal of medical genetics*, *36*(6), 437-446.
- Forsythe, E., Kenny, J., Bacchelli, C., & Beales, P. L. (2018). Managing bardet–biedl syndrome—now and in the future. *Frontiers in pediatrics*, *6*, 23.