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Laurence Moon Bardet Biedl Syndrome: A Case Report

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Abstract:
Children born from consanguineous marriages are more likely to have Laurence Moon Biedl syndrome, an extremely rare autosomal recessive condition. In addition to obesity, limb abnormalities, mental retardation, hypogonadism, and renal illness, it is marked by early-onset retinal degeneration. We describe a 4-year-old boy's case in this report who complained of night blindness. He had retinitis pigmentosa with subsequent optic atrophy, polydactyly, central obesity, and hypogonadism. The patient was identified as having LMBBS.
Key words*: Laurence Moon Bardet Biedl syndrome (LMBBS), Retinal dystrophy, polydactyly.*

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I. Introduction

Laurence-Moon-Bardet-Biedl syndrome is a rare, genetically diversified, autosomal recessive disorganization.

The clinical features of Laurence-Moon-Bardet-Biedl syndrome include early onset night blindness with typical or atypical retinitis pigmentosa, post-axial polydactyly, central obesity, mental retardation, hypogonadism, and structural renal deformity. [1] Ataxia, sensorineural hearing loss, speech impairment or slowness, congenital cardiac issues, and hepatic fibrosis are among more minor disorders. Clinical reflection in the eye includes pigmentary retinopathy, poor visual acuity, and vision loss often ensues due to impaired photoreceptors in retinal tissue with macular involvement leading to night blindness initially, followed by complete blindness in most cases.[2] In patients with an archetypal presentation of LMBBS, truncal obesity is markedly prominent even though the birth weight is usually normal.[3] We present a case with a typical phenotype and the existence of a sister with comparable symptoms.

II. Case History and Examination:

A 4-year-old male youngster born of a consanguineous marriage arrived at the PES Institute of Medical Sciences and Research, Kuppam, with the primary complaints of blurry vision at night and jerky eye movements. Although the patient's older sister had identical symptoms in the past, she wasn't brought in for a physical.

The prenatal, natal, and postnatal phases went without a hitch. Except for a decline in IQ, developmental milestones were met. An examination of the body confirmed the presence of polydactyly in the hands and feet. (See Figure1 and 2). There were symptoms of hypogonadism, including central adiposity, epicanthal inversus (Figure3). Visual acuity was defined as the ability to perceive light in both eyes during an ophthalmological test. Both eyes have posterior subcapsular cataracts, as seen under the slit lamp. Indicating original typical retinitis pigmentosa with subsequent optic atrophy, a fundus examination revealed pallor of the optic disc, bilateral attenuation of vessels, and bone corpuscular pigment (Figure4 and 5).

Figure no 1:Polydactyly in hands

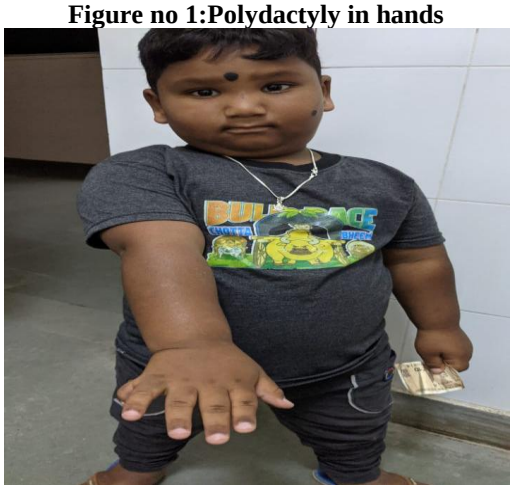
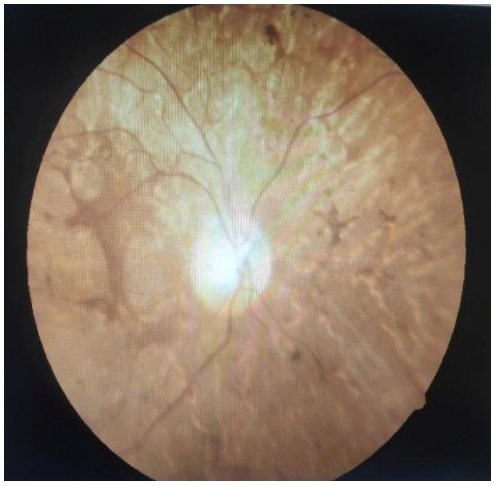


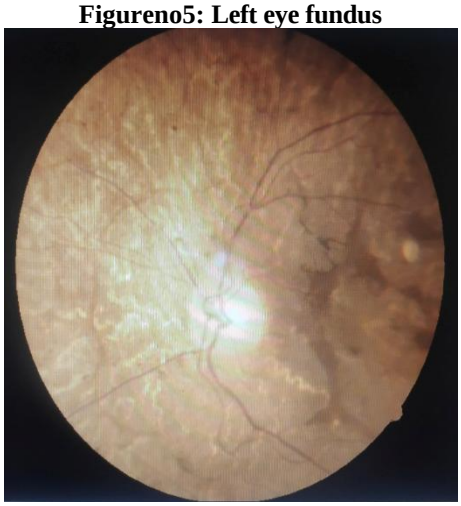
Figure no2:Polydactyly in feet

Figure no 3: Central obesity and epicanthal inversus

Figureno4: Right eye fundus



Figureno5: Left eye fundus



III. Discussion

A 7-year-old female with rod-cone dystrophy, hypogonadism, mental retardation, obesity, and polydactyly was described by Laurence and Moon in 1866. A 4-year-old female patient with rod-cone dystrophy, obesity, polydactyly, and mental impairment was documented by Bardet in 1920. 2. Two years following Bardet's description, Biedl outlined the whole spectrum of clinical indications, including gastrointestinal problems, anal atresia, mental deficit, and anomalies of the skull. [4] Since these discoveries, the condition known as Laurence-Moon-Bardet-Biedl syndrome—which frequently affects kids with normal parents (consanguineous marriage)—has been identified as the presence of the aforementioned manifestations as union (LMBBS).

It's still unclear exactly what biological process leads to BBS. The disease's causative genes, BBS1 through BBS12, have been identified. Since BBS proteins are centrosome components and alter ciliary transport, the condition is included in the category of ciliopathies. [5]

The most frequent main manifestation of BBS is retinal deterioration. Patients typically begin with night blindness and eventually become blind. [6] Night blindness is often diagnosed at a mean age of 8.5 years, but in our instance, it was diagnosed at 4 years old [7]. Obesity typically develops during childhood and gets worse as people age. [8] Brachydactyly and post-axial polydactyly of the hands and feet are two frequent limb malformations. Hypogonadism, mental retardation, and renal failure are other prominent characteristics.

Additionally, other small features have been discovered and described. including speech and language impairment, psychosis, developmental abnormalities of the facial bones, multiple pigmented nevi, hearing loss, diabetes mellitus, cardiovascular abnormalities, dental growth delay with dental abnormalities, gastrointestinal stenosis or atresia, and Hirschsprung's disease. [8]

Aunfavorable prognosis for LMBBS includes early onset vision loss (primarily night blindness), central obesity, hypertension, and diabetes mellitus. LMBBS who have vision problems require low vision aids.

Regular ophthalmological examinations, blood pressure checks, renal function monitoring, and tests for diabetes mellitus and lipid profiles are all part of surveillance.

IV. Conclusion

LMBBS causes significant morbidity and mortality due to its clinical symptoms. With quick evaluation, doctors can more accurately identify and treat this problem, allowing those who are afflicted to live to the fullest and better integrating into society. Since consanguineous marriage is linked to the majority of reported cases of LMBBS, indicating that it is a significant contributor to LMBBS, affected families should receive genetic counselling, especially those with a history of consanguineous marriages. This is done to increase awareness of the possibility of passing on the condition to future generations, as did the patient's elder sister in our case, as well as to get the other family members screened.

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