IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 22, Issue 10 Ser.9 (October. 2023), PP 06-08
www.iosrjournals.org

Laurence Moon Bardet Biedl Syndrome : A Rare Case Report In A Tertiary Care Hospital In South India

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Abstract
Laurence Moon Bardet Beidl Syndrome (LMBBS) is a rare autosomal recessive disorder which involves multiple organs in the body. It is common in children born out of consanguineous marriages and both sexes are equally affected. The main features of this syndrome include progressive cone-rod dystrophy, nystagmus, obesity, polydactyly, mental retardation, learning disabilities, speech disorders, developmental delay, renal abnormalities, polyuria/polydypsia, hypogenitalism, hypospadias, ataxia and poor motor coordination. Here we present a case report of a 32 year old male with complaints of marked diminution of vision in both eyes since more than 20 years and other features suggestive of LMBBS.

Key words: *Retinitis Pigmentosa, Mental retardation, Obesity, Polydactyly, Hypospadias*

Date of Submission: 21-10-2023 Date of Acceptance: 31-10-2023

I. Introduction

Laurence Moon Bardet Biedl Syndrome (LMBBS) is a ciliopathic autosomal recessive genetic disorder which is more common in regions where consanguineous marriages are prevalent. The features are usually apparent within the first 10 years of life, with nyctalopia occuring early [1]. It is rare with the incidence varying from 1:140,000 to 1:160,000 live births in North America and Europe [2] while it is much higher in some Arab countries, approximately 1:13,500 live births [3]. It is extremely uncommon in India with less than 15 cases being reported as of 2009 [4].

A broad spectrum of ocular associations are seen, namely typical or atypical retinitis pigmentosa, rod cone dystrophy, refractive errors like myopia and astigmatism, anisometropia, keratoconus, strabismus, low vision, nystagmus, cataract, optic atrophy, peripheral visual fields constriction, fundus abnormalities like arteriolar attenuation, waxy pallor of disc, bony spicule pigmentation and pigment atrophy [5]. The degenerative changes are rapidly progressive and involve the macula early, causing significant visual impairment and making the patients legally blind by the age of 30 years [6].

Other features of this syndrome include obesity, polydactyly, mental retardation, learning disabilities, speech disorders, developmental delay, renal abnormalities, polyuria/polydypsia, hypogenitalism, hypospadias, ataxia, mild spasticity, diabetes mellitus and poor motor coordination. Less common feautures are diabetes mellitus, hepatic fibrosis, neurological disorder, congenital heart disease, facial dysmorphism and dental anomalies. A wide range of renal anomalies is particularly common, causing significant morbidity and is found to be the leading cause of mortality in patients with LMBBS [8].

II. Case Report

A 32 year old male patient presented to our hospital with complaints of painless progressive marked diminution of vision since he was about 11 years of age. Detailed history revealed he was born out of a second degree consanguineous marriage and has 4 siblings who are asymptomatic. His parents were phenotypically normal and family history was not significant. The prenatal, natal and postnatal period was uneventful. He could manage school upto grade 6 after which he had dropped out due to low vision. Upon clinical examination, he was found to be well oriented to time, place and person and was obese with a BMI of 34.2 kg/m2 (Figure 1). He also had speech difficulties, mild mental retardation, polydactyly in all 4 limbs (Figure 2,3), hypogonadism and hypospadias.

Figure 1

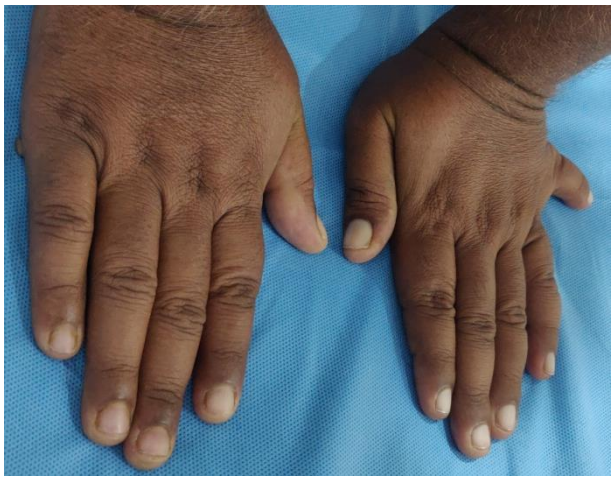

Figure 2

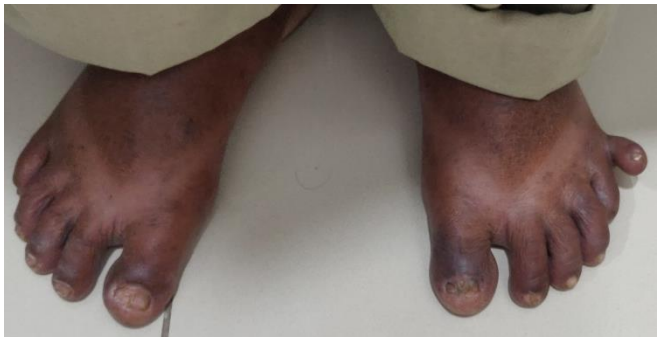

Figure 3

On ophthalmological examination, vision was counting fingers close to face in both eyes. He had strabismus and nystagmus. Slit lamp examination showed presenile immature cataract, NO4 NC4 P4 according to Lens Opacities Classification System (LOCS III) with idiopathic phacodonesis in both eyes. Fundus examination by indirect ophthalmoscopy (IDO) revealed hazy media and waxy pallor of disc, arteriolar attenuation, multiple bony spicule pigments in mid periphery and generalized retinal atrophy consistent with the diagnosis of retinitis pigmentosa. Left eye small incision cataract surgery (SICS) with rigid posterior chamber intraocular lens implantation was done under local (peribulbar) anaesthesia. On post operative day 1, there was no improvement in vision while the cornea was clear and the intraocular lens was stable. Preoperative fundus photo was not clear due to cataract while the postoperative fundus photo of the left eye is as shown in Figure 4.

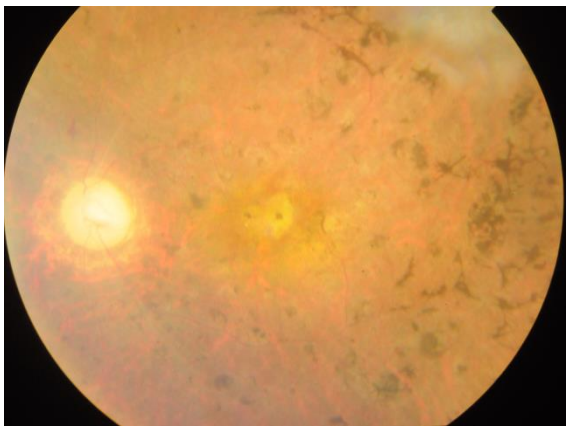

Figure 4

III. Discussion

LMBBS is an uncommon autosomal recessive disorder in which the parents are usually asymptomatic and do not manifest any signs or symptoms but carry the defective gene and pass it on to their offsprings. There is 25% risk of the offsprings being affected by the disorder [5], therfore genetic counselling is of utmost importance along with public education on the potential risks in offsprings born out of consanguineous marriages.

Diagnostic dilemma still exists between Lawrence-Moon syndrome (LMS) and Bardet-Biedl syndrome (BBS). Due to multiple overlapping features, researchers believe BBS to be a part of LMS [9]. The new diagnostic criteria provides guidelines for the diagnosis of LMBBS [10].

Treatment of this condition requires a multimodal approach involving a team comprising an ophthalmologist, paediatrician, physician, orthopedician, audiologist among others. It mainly involves symptomatic treatment and screening for the presence of any associated conditions like diabetes, hypertension, kidney and liver dysfunction. Refractive errors and keratoconus in early stages can be corrected with glasses/contact lens. Frequent consultation with an ophthalmologist is recommended. Low vision aids can particularly be useful in patients with rod cone dystrophy to gain some useful vision to perform their daily activities. However there is no proven effective treatment to either prevent or stop the deterioration in vision.

Due to the uncommon nature of this disorder, diagnosis is often missed. Early diagnosis and adequate treatment by a multidisciplinary team will help patients with this syndrome attain their full potential. This case is being reported due to the rarity of almost all important features of this syndrome being present in our patient.

IV. Conclusion

The disease severity can vary from patient to patient and even within families, significant heterogenicity can be seen. In advanced stages of this condition, there can be considerable morbidity and mortality. An early diagnosis is the key to help patients fare better and therefore it is imperative that all health care professionals are aware of the varied and multisystem presentations of this syndrome. A timely and multidisciplinary management can help these patients lead near normal lives and integrate well into the society. In addition, public education regarding the potential risks of consanguineous marriages and the advantages of genetic counselling should be emphasized.

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