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Document heading **doi: 10.21276/apjhs.2016.3.4.33** **Case report**

Laurence Moon Bardet Biedl Syndrome-A Case Report

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ABSTRACT

Laurence Moon Bardet Biedl Syndrome is a genetic abnormality associated with disorders like retinitis, obesity, polydactyl, learning disability. We report such a case in a 17 year old male patient.

Key words: Uterine contractions Amniotomy**,** Syntocinon

Introduction

Laurence Moon Bardet Biedl Syndrome comprise of Laurence Moon syndrome and Bardet Biedl syndrome. Physicians John Zachariah Laurence and Robert Charles Moon were first to describe this condition in a paper published in 1866. Laurence Moon syndrome shows characteristic findings of Retinitis pigmentosa (Rod-cone dystrophy), hypogenitalism, mental retardation and spastic paresis. Bardet Biedl syndrome shows features of Retinitis pigmentosa, postaxial polydactyly, obesity, learning disabilities and hypogenitalism in males.[1-3]

Case Report

A 17 year old male patient reported to the ophthalmology department, Narayana Medical College, Nellore, Andhra Pradesh, with a chief complaint of loss of vision in dim light since childhood, which slowly progressed over years. He was joined in a blind school at the age of 6 years. He took ophthalmologist opinion and was prescribed spectacles after informing the child and his parents about the night blindness and progressive loss of vision. Every year from then on, ophthalmologist opinion was taken and spectacles with increased power were given. He did not gave any history of double vision or photophobia. He had speech difficulty and slight hearing loss. He gave a positive family history. There was no history of developmental delay and consanguineous marriage. General systemic examination revealed that the patient was obese with Body Mass Index of 29.29 (Grade 1 Obese). Polydactyl of lower limbs was seen (Fig 1). On slit lamp examination, cornea was clear. Fundus examination showed pale, waxy optic disc with attenuation of arterioles and bony corpuscular pigment seen at periphery giving the impression of primary typical retinitis pigmentosa with consecutive optic atrophy (Fig 2). Keeping the observed findings of night blindness, obesity, polydactyl, we gave a diagnosis of Laurence Moon Bardet Biedl Syndrome and prescribed spectacles and Vitamin A supplements. The patient was asked to come for regular follow up.

Discussion

Bardet (1922) first reported Laurence Moon Biedl Bardet syndrome as an autosomal recessive disorder with characteristic features. The patients with this syndrome have key features of pigmentary retinopathy, polydactyly and obesity. Biedl, later added mental retardation and hypogonadism to this syndrome. Other systemic features include short stature, congenital heart

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block, deafness and neurological disorders. Artur Biedl (1922), an Austrian professor of pathology and endocrinology added mental deficiency and genital hypoplasia to this syndrome in their report in 2 siblings.[4, 5]Along with the characteristic features, the patients with this syndrome at times present with other findings like intellectual impairment, cardiovascular abnormalities, deafness and dental anomalies[.4-6]Studies have shown the presence of rod-cone dystrophy in 96% and the average of first notice of night-blindness to be 9 years. Our patient was 17 year old. Polydactyl has been reported in upper or lower limbs. Most of the reported cases were females, whereas our patient was male. Our patient had polydactyl in lower limb. In 75 % cases obesity was found with mean BMI to be 33 kg/m^2. Our patient was also obese with BMI 29.29.5, 7

The features have been categorized into primary and secondary

Primary features: include Rod-cone dystrophy, Polydactyly, Obesity, Learning disabilities, Hypogonadism in males and renal anomalies.

Secondary features: include Speech disorder, Brachydactyly, Developmental delay, Polyuria/polydipsia, Ataxia, Poor coordination/clumsiness, Diabetes mellitus, Left ventricular hypertrophy, Hepatic fibrosis, Spasticity and Hearing loss.

For diagnosis of this syndrome the patients should have either four primary features or three primary and two secondary features. Our patient had 3 primary features (Rod-cone dystrophy, Polydactyly, Obesity) and 2 secondary features (Speech disorder and Hearing loss). Hence we diagnosed the patient to be suffering from Laurence Moon Biedl Bardet syndrome[6-9].Studies have been carried out to find out the biochemical mechanism that leads to this syndrome. It has been thought that twelve genes (BBS1 to BBS12) might be responsible for the disease. As these BBS proteins are components of the centrosome that influence the ciliary transport, this syndrome is categorized under the spectrum of ciliopathies.[7, 10]

Conclusion

Here we present a case report of Laurence Moon Bardet Biedl syndrome in a 17 year old male with family history. He presented with characteristic findings of early onset blindness and ocular findings of Retinitis pigmentosa, optic atrophy. He also presented characteristic general features like obesity, polydactyly etc.

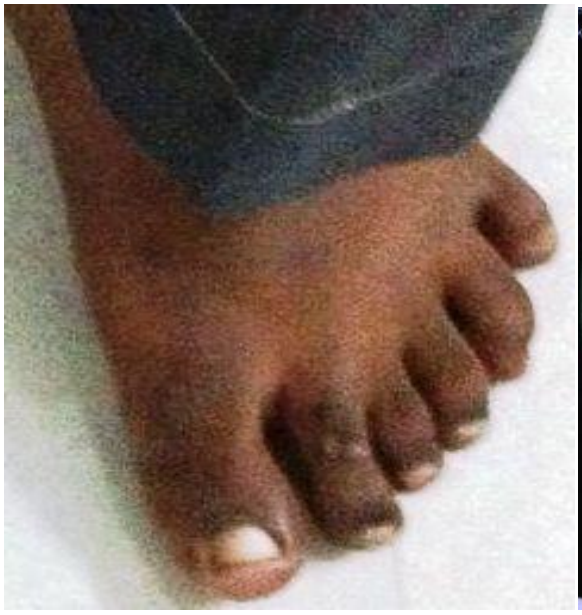

Fig 1:Polydactyl of lower limbs

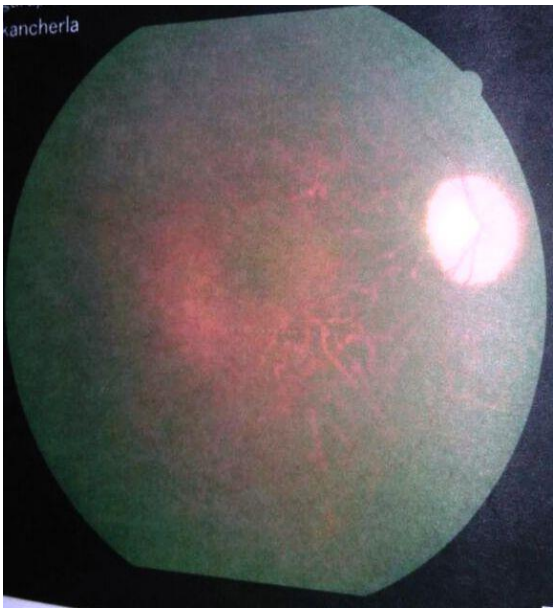

Fig 2:Ocular examination

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