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Case Report

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BARDET BIEDL SYNDROME PRESENTING WITH RETINITIS PIGMENTOSA AND SEIZURES: A RARE CASE REPORT

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ABSTRACT

Bardet biedl syndrome (BBS) is a rare, genetic disorder with involvement of multiple systems and wide spectrum of clinical features. It is also known as Laurence Moon Syndrome (LMS). Principal hallmark of Bardet Biedl Syndrome are red cone dystrophy, obesity, polydactyl, hypogonadism, mental retardation and renal dysfunction. Consanguineous marriage is usually the common cause. Bardet biedl syndrome is inherited mostly as an autosomal recessive trait. It affects males and females evenly. Treatment of Bardet biedl syndrome is directed towards the specific symptoms that are discernible in each individual. We present a case of 35 years male patient with Bardet Biedl Syndrome presenting in medicine department with seizures, moon shaped face, learning difficulties, polydactyly of upper and lower limbs and retinitis pigmentosa. Had a history of Hypertension, Diabetes mellitus and night blindness since childhood. Therefore it is an typical and interesting case report and then the literature is reviewed.

KEYWORDS: Bardet biedl syndrome, obesity, polydactyly, seizures.

INTRODUCTION

Laurence Moon Bardet Biedl Syndrome, first defined by Bardet in 1922 is an rare genetic disorder which is an autosomal recessive disorder characterized by structural and functional abnormalities of organs and tissues with diverse embryonic derivation.[1] Foremost manifestations are retinitis pigmentosa, postaxial polydactyly, central obesity, mental retardation, hypogonadism and renal dysfunction. Other feautures not always present include hepatic fibrosis, diabetes mellitus, neurological, speech and language deficits, facial dysmorphism and developmental delay.[2] Prevalence rate ranges from 1 in 100 000 to 1 in 160 000.[3]

In this case, patient generally has onset of symptoms within the first 10 years of life and associated feautures are poor night vision, Nystagmus and seizures.

CASE REPORT

A 35 year old male patient presented to medicine department with complaints of seizures. On examination he was found to be having retinitis pigmentosa, night blindness since childhood which progressed gradually over years and polydactyly. There was no history of consanguineous marriage between parents. Past history revealed that Diabetes Mellitus since 8 years, Hypertension since 10 years and Epilepsy since 8 months.

Systemic examination revealed obesity, developmental delay and kidney problems. Fundus examination revealed that retinitis. Cardiovascular and urinary systems were normal. Lab investigations including CBC, ESR, electrolytes and urine examinations were normal.

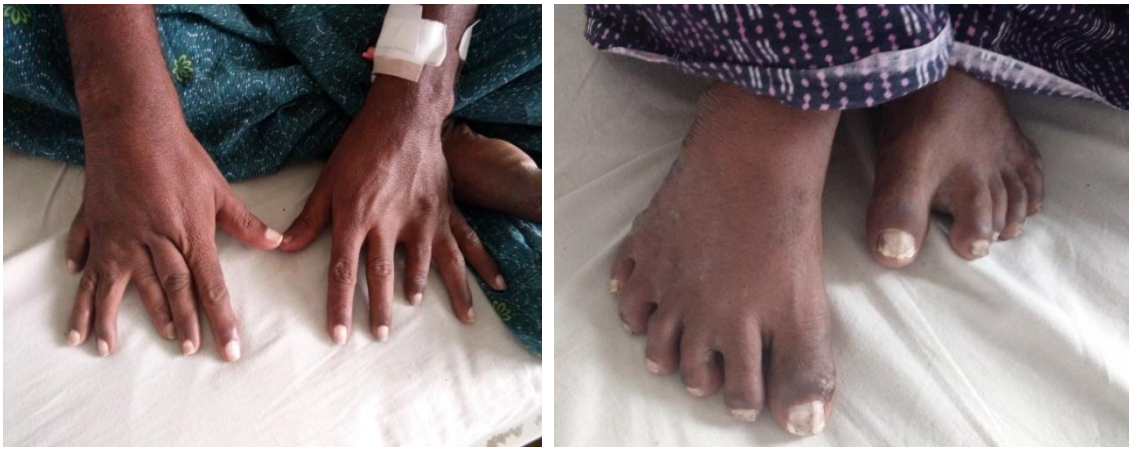

Fig 1: Polydactyly of both upper and lower limbs.

Abdominal and echocardiogram were also normal. Symptomatic treatment was given to patient and managed conservatively for 9 days and then discharged.

DISCUSSION

Bardet (1922) first reported Laurence Moon Biedl Bardet Syndrome as an autosomal recessive disorder with characteristic feautures. The primary clinical feautures include retinitis pigmentosa, polydactyly, central obesity, hypogonadism, cognitive impairment and renal dysfunction. Secondary features include speech disorders or delays, eye abnormalities, ataxia, diabetes mellitus, hepatic fibrosis and congenital heart disease.[4]

Our patient had polydactyly, obesity, retinitis pigmentosa, hypogonadism, mental retardation and diabetes mellitus i.e five primary and one secondary features. Patient generally has onset of symptoms within the first pigmentary changes, bone specular pigmentation and areas of white deposits. Disparate features include Presence of polydactyly of both upper and lower limbs, (fig 1), hearing loss, learning disabilities, moon shaped face and short stature. By above findings diagnosis is confirmed by physician as Laurence Moon Biedl Syndrome. Neurological manifestations are uncommon and rarely seizures may be associated. Seizure was the presenting case, which made this case rare.[4,5]

The treatment of Bardet Biedl Syndrome is directed toward the specific symptoms that are apparent in each individual. Treatment may require the coordinated efforts of a team of a specialists and healthcare professionals. Individuals with Bardet Biedl Syndrome should undergo regular ophthalmologic examinations as well as periodic assessments to determine the presence of complications potentially with the disorder such as kidney dysfunction, diabetes mellitus, liver function and high blood pressure.[3]

Some researchers believe that Bardet Biedl Syndrome is a sub division of Laurence Moon Syndrome. Hence the term Laurence Moon Biedl Syndrome has gained universal acceptance in the world literature.[6,7]

CONCLUSION

We report a typical case of Laurence Moon Bardet Biedl Syndrome in a male of 35 years old. He presented with early onset blindness and other ocular features like Retinitis Pigmentosa. He also showed characteristic general features of polydactyly, obesity, cognitive impairment and hearing loss. He also had kidney problems and developmental delay. Seizures is a rare association with Bardet Biedl Syndrome.

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