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MENTAL RETARDATION WITH LAURENCE MOON BARDET BIEDL SYNDROME (LMBBS)

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ABSTRACT

As we all know Laurence Moon Bardet Biedl Syndrome (LMBBS) is a rare autosomal recessive disorder, mostly seen in children born from grade II or grade III consanguineous marriages. It is described by early onset retinal dystrophy, central obesity, forelimb malformation, mental retardation, renal dysgenesis, hypogonadism and abnormal organ development. In this case report we dispensing a 8 year old boy who presented with complains of night blindness. On examination he had polydactyly, mental retardation, obesity, hypogonadism, microdontia and retinal dystrophy with no hearing impaired. The patient was diagnosed with LMBBS.

INTRODUCTION

Laurence-Moon-Bardet-Biedl Syndrome is a infrequent, genetically diversified, autosomal recessive disorganization presented early with clinical features like early onset night blindness with typical or atypical retinitis pigmentosa, post axial polydactyly (extra figure either in forelimb or hind limb), central obesity, mental retardation (MR), hypogonadism and structural renal deformity.[1]

Other minor defects include speech hamper, slurred speech or slow down, sensorineural hearing loss (SNHL), ataxia, congenital heart problems as well as hepatic fibrosis. Here we report a case with typical phenotype and presence of similar complaints in a sibling.

Case History and Examination

A 8 years old male presented with chief complaints of gradual painless blurring of vision along with nyctalopia from both eyes. The child patient, which is a product of non-consanguineous marriage, presented to the ophthalmology outpatient department (OPD) of Shahu Maharaj Lions Pride Eye Hospital, Ichalkaranji, and Maharashtra. His parents gave history of similar complaints in the elder sister of the patient, which is history of night blindness. She was not brought for examination in our clinic. Past history of Antenatal, natal and postnatal periods were uneventful. Developmental milestones were achieved better with increase in age except for decreased intelligence as well as grasping (recall) power.

Physical examination revealed presence of polydactyly and brachydactyly (short digits) of both hands and feet. (Photograph A and D).There was central obesity (Photograph B), dental anomalies (Image C) features of hypogonadism and micro penis. Central nervous system examination and chest examination was non significant. Well co operative with time, place and person. Chest shows air entry bilateral equal with no abnormal vesicular sounds. Muscle tone, power or rigidity within normal limit. Per abdomen examination were no significant.

On ophthalmological examination, visual acuity was 6/60 in both eyes. Conjunctiva, cornea within normal limit and normal depth of anterior chamber.Van Herick grade 4. Extraocular movements were full and free including duction and versions. Fundus examination revealed pallor of optic disc, bilateral attenuation of vessels, decrease in artery and vein ratio with retinal pigmentary changes mostly outer retina could be bony specules in mid periphery.

The diminution of vision was not accompanying by metamorphopsia or micropsia. He didn't give history of redness or discharge. There was no history of ocular pain associated with eye movement. Anterior segment finding was normal in both eyes. On the basis of above finding, diagnosis of Laurence-Moon-Bardet-Biedl Syndrome which is association of retinitis pigmentosa was made. Patient was treated with oral Vitamin A 2 lakh IU tablet, vitamin E 400 IU. Rehabilitation and low vision aids (LVA).



Image (Photograph) A: Shows Polydactyly of Right Hand



Image (Photograph) B: Shows Central Obesity with Fat Deposition



Image (Photograph) C: Shows Abnormal Dental Development



Image (Photograph) D: Shows Extra Fingers or Digit in Both Feet (Polydactyly)

DISCUSSION

In 1866, Laurence and Moon explained a case of 7-year-old female with rod-cone dystrophy, hypogonadism, mental retardation, obesity and polydactyly. In 1920, Bardet described a 4-year-old female patient presented with rod-cone dystrophy, obesity, polydactyly and mental retardation. [2] Two years after Bardet's report , Biedl highlighted the complete scenario of clinical signs which includes skull abnormalities, anal atresia, mental deficiency and gastro-intestinal disorders.[2] Since these discoveries' presence of above manifestations as union, often in children with normal mother and father (consanguineous marriage) has been called as Laurence-Moon-Bardet-Biedl syndrome (LMBBS).

The detailed biochemical process that causes LMBBS is still ambiguous. Twelve genes (BBS1to BBS12) that are responsible for the disease have been identified. BBS proteins are components of centrosomes and affect the ciliary transport, hence the disease falls under the spectrum of ciliopathies.[3] Retinal dystrophy is most common major manifestation present in LMBBS. Patients usually present with night blindness which gradually progresses to blindness. [4] Usually, night blindness is noticed at the mean age of 8.5 years but in our case, it was noticed earlier at the age of 6 years as history given by parents .[5] Obesity usually begins in childhood and progresses with age.[6] Limb abnormalities like post axial polydactyly, brachydactyly (short digits) of both hands and feet are common. Other major features include hypogonadism, mental retardation and renal dysfunction.

Multiple minor features have also been found and documented. Which consist delayed developmental milestones with growth ,speech as well as language short fall, psychosis, facial bones abnormality in developmental form, multiple pigmented nevi, hearing impaired, maturity onset diabetes of the young (MODY), cardiovascular abnormality, dental growth delay with dental abnormality, gastrointestinal stenosis or atresia and Hirschsprung's disease. [7] Female manifestations in LMBBS include hypoplastic fallopian tubes, uterus, ovaries, vaginal atresia, no vaginal opening with absent urethral orifice. Renal failure is common etiology in BBS patients, renal symptoms consist renal calculi, chronic renal failure, parenchymal cysts, calyceal clubbing and scarring. The mentioned features were absent in our case report.[8]

LMBBS with visual impairment need low vision aids (LVA). Surveillance includes regular ophthalmological examination, blood pressure measurement, monitoring of renal function, and regular testing of lipid profile and sugar to rule out diabetes mellitus.

CONCLUSION

Diagnosis of Bardet Biedl syndrome is mainly clinical as genotyping is not available in many places especially in developing countries like India and Nepal. Multidisciplinary approach is required for management, to provide good prognosis and quality of life to our patients.

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