LAURENCE MOON BARDET BIELD SYNDROME a case report
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Abstract: Laurence moon Bardet biedl syndrome is a rare congenital syndrome with an incidence between 1 in 160,000 and 1 in 15,000, commonly diagnosed with the pentad of mental retardation, pigmentary retinopathy, polydactyly, obesity, and hypogonadism. We report one such case.

Case report: A 19 year old boy was brought with complaint of excessive weight gain noticed since early childhood. He was born out of non-consanguinous marriage at term, with no perinatal complications. The patient was the last child of his family which includes three elder sisters. All other family members were normal. He had progressive deterioration in vision for the past six years, starting with difficulty in seeing at night. There was history of developmental delay also. His mother noticed that his penis was very small. He had dropped out of school at seven years of age. At present he does his daily activities with the help of his mother. He also had a giant nevus over his face.

Examination: He was markedly obese with predominant fat distribution over face, chest, abdomen and neck. His weight was 85 kg (+4.6 SDS), height was 156 cm and BMI was 35. His blood pressure was 116/70 mm Hg. There was postaxial polydactyly of all four limbs. He had a total of 24 digits. Examination of the external genitalia revealed stretched penile length 2.8 cm (microphallus). Testes was normal in volume. Secondary sexual characters were minimally present. Ophthalmologic examination revealed visual acuity of 2/60 in both eyes. Retinoscopy revealed high myopia and astigmatism. Fundus showed arterial attenuation and bony corpuscles in mid peripheral region suggestive of retinitis pigmentosa.

INVESTIGATIONS: Random blood glucose was 84 mg%, liver function tests were normal. Blood urea was 47 mgs% and creatinine was 1.8 mgs%. The lipid profile was as follows: Total cholesterol 178 mgs%, H.D.L.: 41 mgs%, V.L.D.L.: 24 mgs%, L.D.L.: 107 mgs%, Triglycerides 117 mgs%; all of which are normal. LH, FSH and testosterone were within normal limits. Urine examination revealed +1 proteinuria. 24-h urinary collection revealed 0.18g of protein. Ultrasound scan of abdomen showed fatty, enlarged liver with normal kidneys. ECG and ECHO revealed left ventricular hypertrophy. Diagnosis of Laurence-Moon-Bardet-Biedl syndrome having central obesity, postaxial polydactyly, mental retardation, retinitis pigmentosa, hypogonadism and functional renal impairment was made. He was prescribed glasses, advised for fat and refined carbohydrate-restricted diet and exercise and advised follow-up after 3 months.

DISCUSSION: The patient above is a classical case of Laurence Moon-Bardet-Biedl syndrome. The syndrome was independently described by Bardet and Biedl in the 1920s with the classic pentad polydactyly mental retardation, hypogonadism, mental retardation, retinitis pigmentosa and also renal failure along with other clinical manifestations. In 1865 Laurence and Moon described a similar syndrome in 4 members of a family which along with the cardinal features had spastic paraparesis which was not reported by bardet and biedl! The prevalence is 1: 160 00 to 1:15000. Common genes affected include BBS1, BBS2, ARL6/BBS3, BBS4, BBS5, MKKS/BBS6, BBS7, TTC8/BBS8, B1/BBS9, BBS10, and TRIM32/BBS11. The syndrome is transmitted as an autosomal recessive trait. There is considerable heterogeneity with sporadic mutations and variable expression. Majority of the patients are born out of nonconsanguinous marriages. Hence sporadic mutation is also said to have a significant effect along with summation of multiple gene defects in families. The bodily changes are the result of a dysplasia of two kinds of primitive tissues: (1) The primitive mesenchyme changes which result in the skeletal defects as polydactyly and syndactyly, and (2) epiblastic tissue, which results in changes of diencephalon. It is primarily considered as a ciliopathy with the genes causing ciliary dysfunction resulting in varied clinical presentation. The exact pathogenetic sequences haven’t been clearly established.

DIAGNOSTIC CRITERIA: It is established by the clinical criteria suggested by Beales et al with the presence of four primary features or three primary features plus two secondary features.
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, left ventricular hypertrophy, diabetes mellitus, hepatic fibrosis, and spasticity.

**PROGNOSIS:** This syndrome has an adverse prognosis, with early onset of blindness, obesity, hypertension, and diabetes mellitus. Renal impairment is frequent and often goes undetected. This is significant in that early death often occurs because of the renal disease.

**TREATMENT** visual impairment need visual aids and educational programs. For management of obesity, diet, exercise, and behavioral therapies are advocated. Accessory digits may be removed surgically to prevent functional interference and poor fitting of footwear. Early intervention and special education is needed to address cognitive disability. Speech delay/impairment needs early diagnosis and speech therapy. Hormone replacement therapy is advocated to correct hypogonadism. Renal anomalies and hypertension are treated as in the other affected children.

**CONCLUSION** Laurence moon syndrome and Bardet-Biedl syndrome are considered as a spectrum of a single disorder. Our case has the five cardinal features of polydactyly, central obesity, hypogonadism, mental retardation, and retinitis pigmentosa along with raised renal parameters, left ventricle hypertrophy, clumsiness, developmental delay, brachydactyly, and astigmatism.

**References**