Original Research Paper

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Medical Science

A RARE CASE OF LAURENCE–MOON BARDET – BIEDL SYNDROME

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ABSTRACT

METHODS : A 15 years old female was brought with chief complaints of diminution of vision both eyes since birth. On ocular examination patient had torsional nystagmus of low amplitude and moderate frequency. Rest of the anterior segment was within normal limits. Fundus examination revealed normal optic disc with few bony spicules in mid peripheral retina with macular lesions and atrophy of RPE in both eyes

RESULTS : On general examination patient had Mental retardation and polydactyly in all 4 limbs. BMI was 32kg/m2 suggestive of obesity. There was history of delayed attainment of milestones with gross developmental delay. Patient gives no history of attainment of menarche . Urine routine and microscopy was within normal limits. USG abdomen was suggestive of hypoplastic uterus. Patient was diagnosed with Laurence–Moon–Bardet–Biedl syndrome. No treatment is currently known for the retinal degeneration associated with this syndrome

KEYWORDS :

INTRODUCTION

One of the most common syndromic retinal degenerations. The cardinal features of BBS are retinal degeneration, genito-urinary tract anomalies, polydactyly, obesity, and cognitive impairment. [1] In some, visual impairment may be severe and onset early, as in LCA. In others with BBS, the retinal degeneration is recognized later in childhood when the child presents in renal failure. Progressive loss of acuity and retinal sensitivity are inevitable; the courses are variable.

CASE REPORT

A 15 years old female was brought with chief complaints of diminution of vision both eyes since birth. On ocular examination patient had torsional nystagmus of low amplitude and moderate frequency. Rest of the anterior segment was within normal limits. Fundus examination revealed waxy pallor disc with few bony spicules in mid peripheral retina with macular lesions and atrophy of RPE in both eyes.

DIAGNOSIS & RESULTS

Using the diagnostic criteria proposed by Schachat and Maumenee, which require the presence of four out of five principal features (retinal degeneration, mental retardation, obesity, polydactyly, hypogonadism), the BBS status of the respondent (affected versus unaffected) was determined. [2] It has been suggested previously that obesity is an indication of obligate carrier status in BBS. [3,4]

On general examination patient had mental retardation and polydactyly in all 4 limbs BMI was 32kg/m2 suggestive of obesity. There was history of delayed attainment of milestones with gross developmental delay. Patient gives no history of attainment of menarche. Urine routine and microscopy was within normal limits. USG abdomen was suggestive of hypoplastic uterus.

CONCLUSION

Patient was diagnosed with Laurence–Moon–Bardet–Biedl syndrome. No treatment is currently known for the retinal degeneration associated with this syndrome.

Figure - 1- Retinal Degeneration

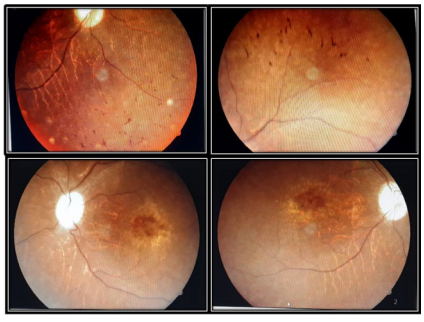


Figure-2 - shows polydactyly

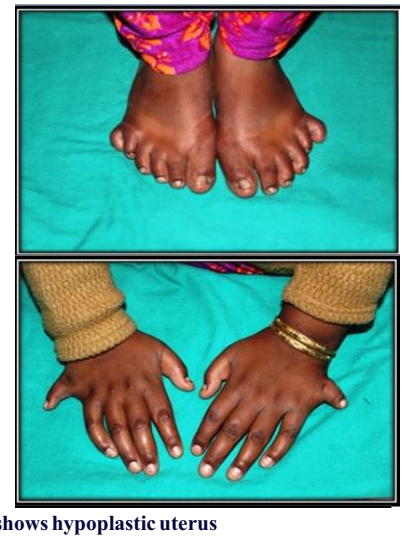
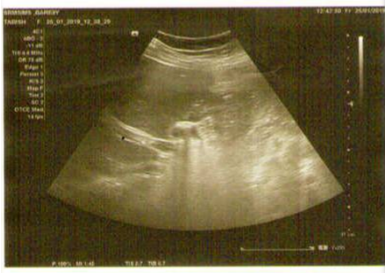


Figure-3 shows hypoplastic uterus

Figure-3 shows hypoplastic uterus



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