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LAURENCE MOON BIEDL SYNDROME IN A YOUNG GIRL WITH OVERLAPPING FEATURES OF TURNER SYNDROME: A CASE REPORT.

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ABSTRACT:

The author presents a case of Laurence Moon Biedl syndrome. A 14 year female was diagnosed as Laurence Moon Biedl syndrome on the basis of polydyctyly, central obesity, mental retardation, delayed puberty (hypogonadism) and primary retinal dystrophy. A novelty in the case of young girl was presence of low occipital hair line and cubitus valgus. Renal impairment was not seen in her case.

INTRODUCTION:

Laurence Moon Biedl syndrome, a rare, genetically heterogeneous autosomal recessive disorder, is characterized by familial occurrence of primary retinal dystrophy, polydactyly, obesity, hypogonadism and mental retardation [1, 2]. For many years the etiology of the Laurence Moon Biedl syndrome has been a question of very unsatisfactory debate. Its establishment as a diagnostic entity, however, is quite secure with over 200 cases recorded in the literature. In 1866, Laurence and Moon reported 4 cases of retinitis pigmentosa occurring in the same family, associated with other unusual abnormalities including hypogenitalism, obesity, mental deficiency and dwarfism. The symptom complex, although observed occasionally by clinicians in the interim, was not recognized as a disease entity until 1920 when Bardet reported an isolated case of polydactyly, adiposogenitalism, and retinitis pigmentosa. French authors are naturally inclined to associate his name with the syndrome. Biedl, in 1922, reported 2 cases in the same family, and he considered that the syndrome in its complete form would include any of such congenital malformations as polydactyly, retinitis pigmentosa, skull deformities, and anal atresia, as well as mental deficiency, obesity, possible genital hypoplasia, and digestive disturbances [3]. Beales and colleagues carried out the largest survey on this disorder, defined the frequency of various manifestations and revised the diagnostic criteria. According to this study, postaxial polydactyly had been present in 69% of patients at birth, but obesity had only begun to develop at around 2-3 years, and retinal degeneration had not become apparent until a mean age of 8.5 years. The study identified some novel clinical features, including neurological, speech, and language deficits, behavioural traits, facial dysmorphism, and dental anomalies [4]. According to another study, all 28 patients examined had severe retinal dystrophy, but only 2 had typical retinitis pigmentosa. Polydactyly was present in 18 of 31 patients, but syndactyly, brachydactyly, or both were present in all. Obesity was present in all but 1 of 25 patients. Only 13 of 32 patients were considered mentally retarded. All 12 women studied had menstrual irregularities, and 3 had low serum estrogen levels (1 of these had hypogonadotropism, and 2 had primary gonadal failure). The remaining women who were of reproductive age had endocrinologic evidence of reproductive dysfunction [5]. Genetic basis underlying this disorder are complex and many experimental animal model studies [6-10] are underway to elucidate all those. Leppert et al [11] advocates heterogeneous linkage of this syndrome to two genetic markers on chromosome 11q, *PYGM* and AFM164zf12 (*D11S913*). In the last, the syndrome keeps a bad prognosis. Patients are generally lost due to renal insufficiency at young ages [12].

CASE DETAILS:

The author presents a very interesting case of Laurence Moon Biedl syndrome in a 14 years old female Mussarat Bibi from a far off village in District Lodhran shown in Figure 1. She presented to medical outpatient department of Bahawal Victoria Hospital with the history of obesity, low general intelligence, polydyctyly and brachydyctyly in right and left foot respectively as shown in Figure 2, and decreased vision since childhood. History

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of primary amenorrhea was also present. Actually the parents belong to poor socioeconomic status and illiterate background. Despite the fact, they had two consecutive issues (elder boy and younger girl) with the above mentioned set of abnormal characteristics, never sought medical advice. It was the death of the elder brother of the patient, few months back which compelled her parents to rush to a medical facility. The patient was admitted to Medical Unit II for complete work up. General physical examination revealed central type of obesity, short stature in accordance with age and gender of the patient, cubitus valgus, low occipital hair line and normally shaped external genitalia. It was note worthy that no pubic or axillary hair growth was appreciated and breast development was also delayed. On nervous system examination, the only positive finding was reduced near and far vision. Cardiovascular examination showed normal and regular pulse with no radio-radial or radio-femoral delay. Blood pressure in all four limbs was 110/70 mmHg with no postural drop. On the day 1 of inpatient assessment, the author made four possibilities in mind, Laurence Moon Biedl syndrome, Psudopsudohypoparathyroidism, Turner syndrome and hypothyroidism, sent routine laboratory and radiological investigations. Her blood complete picture, serum sugar, liver and kidney function tests were within normal range. Serum Calcium, Phosphorus, Alkaline Phosphatase, Parathormone, Thyrorid Stimulating Hormone (TSH) and free serum T3 levels were also within normal limits. Chest X-ray and electrocardiogram were also unremarkable. Pelvic and abdominal sonography was done to see the status of internal genitalia to include or rule out the possibility of Turner syndrome. It shows normal ovaries bilaterally and thick walled uterus producing dense echogenicity. Echocardiography showed normal cardiac chambers with good LV function. Coarctation of Aorta, an association of Turner' was also ruled out. For detailed visual examination, the patient was sent to Consultant Ophthalmologist who reported primary retinal degeneration on fundoscopy. Rest of the ophthalmic examination was unrevealing. The most unfortunate aspect was that the parents were counseled about the nature of hereditary rare disorder and its bad prognosis in very humane and humble manner. The patient and her parents were advised about regular follow up for renal profile. Nothing else could be done!

FIGURE 1:
A 14 YEAR FEMALE: OBESITY, SHORT STATURE AND CUBITUS VALGUS ARE EVIDENT.

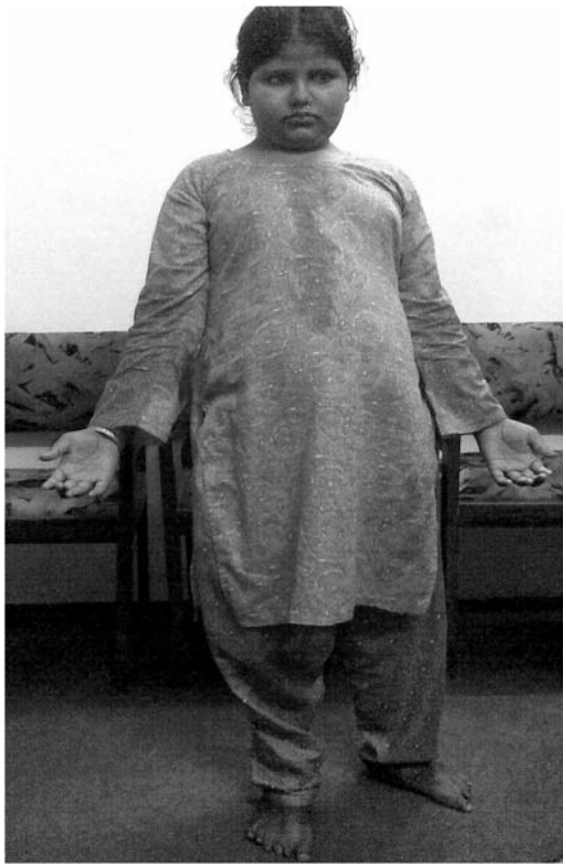


FIGURE 2:
POLYDYCTYLY IN RIGHT FOOT AND BRACHYDYCTYLY IN LEFT FOOT.



CONCLUSION:

To summarize, diagnosis of Laurence Moon Biedl syndrome was made on the basis of polydyctyly, central obesity, mental retardation, delayed puberty(hypogonadism) and primary retinal dystrophy. A noval thing in the case was presence of low occipital hair line and cubitus valgus. Renal impairment

was not seen in this case.

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