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RENAL INVOLVEMENT IN THE LAURENCE-MOON-BIEDL SYNDROME: REPORT OF A CASE AND A SIBLING*

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The Laurence-Moon-Biedl syndrome (LMB) whose salient features are a specific combination of retinitis pigmentosa, hypogonadism, obesity, mental retardation and polydactyly is a rare disorder which is inherited as an autosomal recessive trait and characterized by familial occurrence. The primary defect and the actual prevalence are unknown[1-5]. Over 400 cases have been reported in the literature to date and the incidence is estimated to be about 1 per 160,000. According to some recent reports mortality regarding this syndrome has been found to be a result of terminal renal failure and renal anomalies and these findings must be considered to be the sixth cardinal component of the Laurence-Moon-Biedl syndrome[2,4,6].

A case of the Laurence-Moon-Biedl syndrome with renal involvement and his female sibling is presented in this paper.

Case Reports

Case 1

A twenty-seven-year-old male presented with edema of the legs, trunk and eyelids. His history revealed that he had been delivered after an uncomplicated pregnancy. His sight and hearing were abnormal and had worsened progressively since his childhood. He has been married for three years and has no children. Fifteen days prior the patient noticed edema of the eyelids which was prominent in the mornings and subsided in the evenings. Pollakiuria, nocturia and polyuria have been present for six years. Physical examination revealed a micropenis, small testes, microscrotum, mental retardation, photophobia, vertical and horizontal

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nystagmus, truncal obesity, and gynecomastia. He was able to count the number of fingers from a distance of 50 cm. He also had a high cleft palate and pitting edema of the legs.

Laboratory studies revealed isosthenuric urine and proteinuria (0.5 g/L/24 hr). Thyroid function tests, a hemogram, buccal smear, chromosome analyses, ultrasonography of the liver and spleen, and skeletal roentgenograms were normal. Serum FSH: 15.7 mlu/ml (normal values 2-10 mlu/ml), LH: 24 mlu/ml (normal values 2-15 mlu/ml) and creatine clearance was 55.5 cc/min. The renal echo pattern was slightly increased on ultrasonography. Intravenous pyelography illustrated both kidneys to be smaller than normal displaying irregular contours and calicectasis; the excretion of urographic contrast medium was delayed. Fundoscopy revealed superficial retinal hemorrages and retinitis pigmentosa. Pulmonary function tests showed restrictive and obstructive impairment. The IQ was low. CAT scan of the brain revealed cortical atrophy (Fig. 1). A renal biopsy was performed, Histologically, hyalinization in the glomeruli, thickening of the basal membrane and mesangial proliferation were detected.

Case 2

The thirteen-year-old female sibling of Case 1 also had retinitis pigmentosa, mental retardation, truncal obesity, hypogonadism and minimal hearing loss. Urinalysis revealed two to three red blood cells per high-power field; urine culture and cytology were negative, serum creatinine was 1.4 mg/dl, and creatinine clearance 65 ml/min. Excretory urography (IVP) showed bilateral small, poorly functioning kidneys with calicectasis. Cystoscopy was unremarkable, except for cystitis cystitica. A CAT scan of the brain revealed cortical atrophy.

Discussion

The pathogenesis of this rare syndrome is unknown and of the cases reported most are of the incomplete form. The pentad of the syndrome is present in only 40-45 percent of the cases. If renal involvement is included, then in addition to the five salient features of the syndrome, the six major and many minor components of this disorder such as nystagmus, deafness, skull defects, congenital heart disease, cataracts, atrophy, coloboma of the iris and more rarely ataxia and diabetes mellitus[5,7,8] may be seen in the clinics.

Most of the reported adult cases in the literature are male patients who present with infertility. The pathogenesis of hypogonadism is unknown. Target insensitivity to gonadal hormones, primary end organ failure or hypothalamic dysfunction have been suggested[9,10]. FSH and LH levels were demonstrated to be high in our case supporting primary end organ failure or target insensitivity to gonadal hormones. A CAT scan of the brain revealed cortical atrophy and a normal pituitary gland.

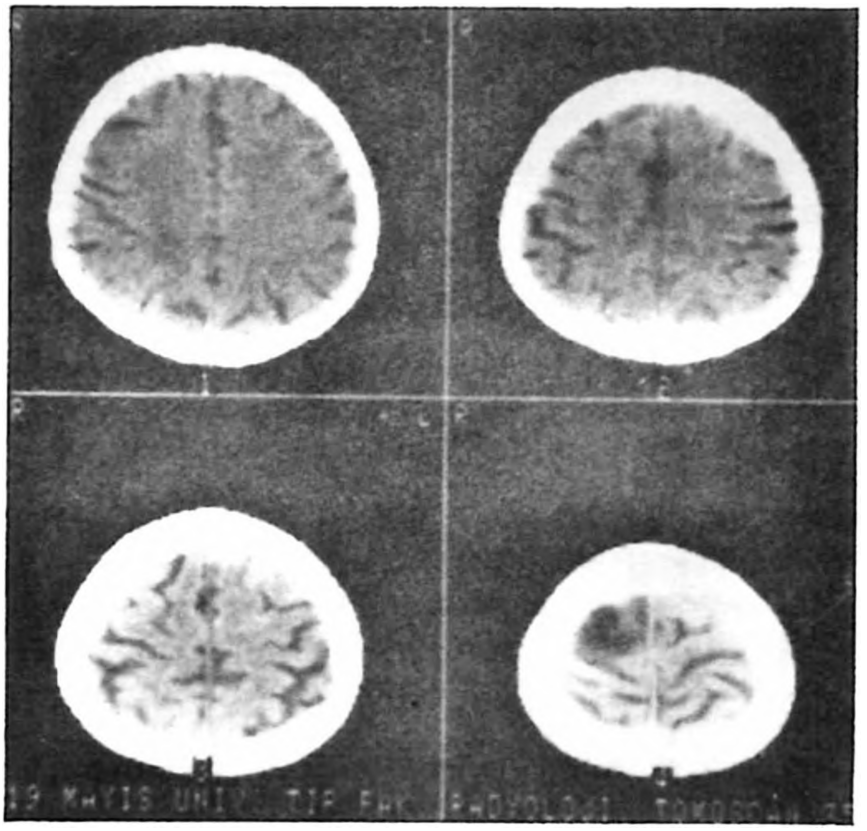

Fig. 1: CAT Scan.

Renal involvement has been reported in 30 percent of the cases of the Laurence-Moon-Biedl syndrome[2]. Recently, more cases have been reported and it is suggested that chronic renal failure and uremia are the principal causes of death in patients with this syndrome. Renal involvement should be regarded as the sixth cardinal finding[1-5]. Renal function tests and renal biopsies of these patients have revealed reduced GFR, reduced maximal concentrating capacity, tubular defects, isolated aminoaciduria and lesions with calicine and tubulo-interstitial cyst formation[2,6]. Glomerular hyalinization and mesangial proliferation which were found in our case has also been reported previously[4,6]. The unknown etiology of mental retardation which is a cardinal symptom of the Laurence-Moon-Biedl syndrome might be the result of cerebral cortical atrophy, a finding seen in our case.

In conclusion, uremia is stated to be the cause of death in more than one-third of the reported cases of LMB. Therefore, in addition to the detection of mental retardation and hypogonadism, renal function should be determined. The pathogenesis of impaired renal function can not be elucidated, but nephronophithisis may be a possible explanation[11].

Summary

A twenty-seven-year-old male who was hospitalized because of renal disease and his thirteen-year-old sister, both having the Laurence-Moon-Biedl syndrome, are presented and the related literature is reviewed for the presence of renal disorders in regard to this syndrome.

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