Bardet Biedl syndrome: a case report

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Abstract

Bardet-Biedl syndrome is rare genetic disorder, characterized by gross physical abnormalities like postaxial polydactyly or syndactyly, obesity, visual disturbances, mental retardation, hypogonadism. Diagnosis based on a group of clinical features. Here I am reporting a case of 14 years old boy presenting with obesity, difficulty in vision and hypogonadism. Bardet-Biedl syndrome was diagnosed and appropriate counselling and symptomatic treatment was discussed with his parents.

Key words: Bardet-Biedl syndrome, obesity.

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Introduction

The Bardet-Biedl syndrome (BBS) is a rare ciliopathic human autosomal-recessive disorder, characterized by cardinal symptoms of marked central obesity, retinal dystrophy, polydactyly, mental retardation and hypogonadism and renal dysfunction.[1] Its prevalence varies from 1:160,000 to 1:13,500, respectively, in northern Europe and in some communities in Kuwait.[2] Higher incidence in Arab populations can be due to the fact of marriage being usual between relatives.[3] BBS is part of a group of human genetic disorders of cilia function, and mutations of 17 genes are reported to be responsible for more than 80% of clinically diagnosed cases.[4]

The diagnosis of Bardet-Biedl syndrome (BBS) is established by clinical findings. Beales et al have suggested that the presence of four primary features or three primary features plus two secondary features is diagnostic. Primary features are: Rod-cone dystrophy, Polydactyly,Obesity, Genital anomalies, Renal anomalies, Learning difficulties and Secondary features are : Speech delay, Developmental delay, Diabetes mellitus, Dental anomalies,Congenital heart disease, Brachydactyly/ syndactyly, Ataxia/ poor coordination, Anosmia/hyposmia. Four primary features or three primary features and two secondary features are required for a clinical diagnosis of Bardet Biedl syndrome.[5,6]

Case reports

Abdur Rashid, 14 years old boy, non diabetic, normotensive hailing from Bagmara,Rajshahi was admitted in RMCH on 01.01.2016 through emergency with the complaints of Difficulty in vision and Progressive weight gain for 6 years. According to the statement of the mother of the patient, he was reasonably well 7 years back then he developed difficulty in vision both near and distant. She also complains of difficulty in vision at night. There was no history of pain, watering or redness of eyes. She also complains of progressive weight gain for the last 6 years.She gives a history of excess food intake with increase appetite. There was no history of cold intolerance, proximal muscle weakness, headache, constipation, excessive sleep, taking any drugs like steroids. None of the family members were suffering from such type of illness. On examination, patientwas obese involving more on central part of the body.Face is rounded but not plethoric. Height-150 cm, weight-91 kg, BMI-40 kg/m2. No anemia, jaundice, clubbing, koilonychia, leukonychia or oedema. No thyromegaly or lymphadenopathy.There are multiple linear striae involving abdomen, thigh and shoulder. No bruise present. No proximal myopathy. Both breasts are enlarged,soft and non tender(bilateral lipomastia). Postaxial Polydactyly present on both hands and feet.Pulse-80b/min,BP-120/80 mmHg,respiratory rate-14/min, temperature-98 F.

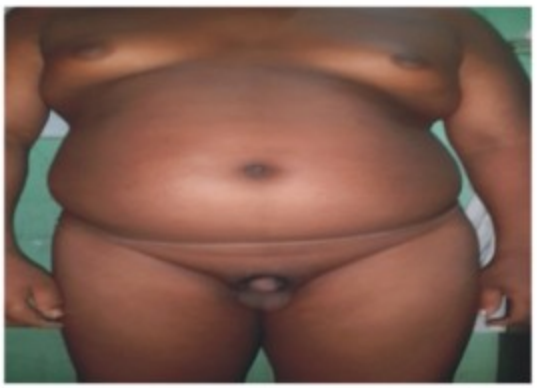

Figure 1: Obesity and small penis.

Higher psychic function revealed normal.Visual acuity reduced to finger counting.Bilateral reduction of temporal field of vision.Color vision normal.Fundoscopy revealed arteriolar narrowing, perivascular bone specule pigmentation and waxy optic disc pallor.Other cranial nerves were normal.Motor and sensory functions were intact. Abdomen distended, flanks full, soft, nontender, bowel sound present, hernial orifices intact. Bilateral small testes (4ml on orchidometer bilaterally) with small external genitalia (4cm) . His serum testosterone was 2.78 nmol/l (lab ref. level of serum testosterone of 12-15 yearsboy : 3.46-11 nmol/l). Other lab investigation reports CBC, RBS, Liver and Renal function tests were normal. Chest X-ray P/A view and Ultrasonography of abdomen reveal no abnormalities. Thyroid hormones and ECG report were also normal. Our patient had four primary features of Bardet-Biedl syndrome retinal changes, obesity, hypogonadism and polydactyly as suggested by Beales et all.[5,6]

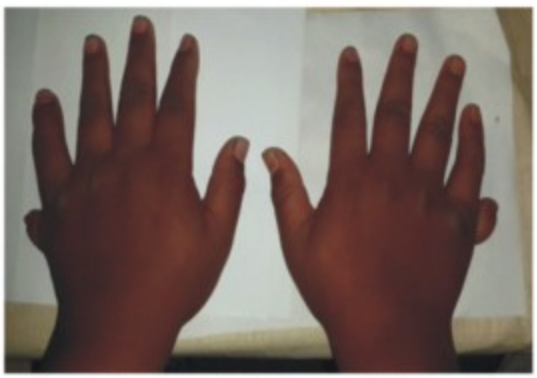

Figure 2: Polydactyly involving hands

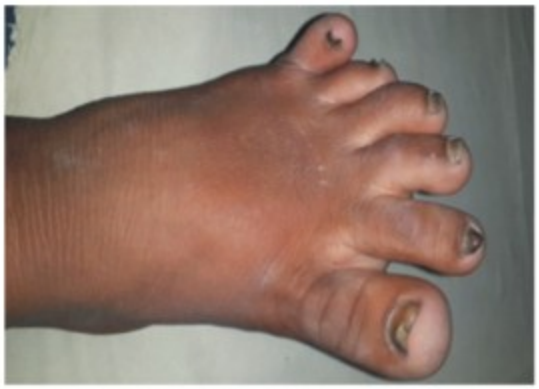

Figure 3 : Polydactyly involving leg

Discussion

Laurence and Moon in 1866 at the Ophthalmic Hospital in South London first described LaurenceMoonBiedlBardet syndrome, but now a days BardetBiedl syndrome and LaurenceMoon syndrome is usually considered a separate entity.[7] In 1920 Bardet described the three of the five cardinal features of the syndrome, polydactyly, obesity and pigmentary retinopathy.[8] LaterBiedl in 1922 added mental deficiency and genital hypoplasia. In 1925, Solis-Cohen and Weiss coined the name 'LaurenceMoonBiedlBardet syndrome'.[8]

Major clinical features are pigmentary retinopathy, poor visual acuity, low vision, and/or blindness caused by an impaired photoreceptor transport mechanism in the retina.[8] Loss of, or reduced sense of, smell (anosmia). Some patients claim extra-sensitive sense of smell.[9] Polydactyly (extra digits) or syndactyly (webbing of fingers and toes). Hypertrophy of interventricular septum and left ventricle and dilated cardiomyopathy. Hypogonadism, renal failure, urogenital sinuses, ectopic urethra, uterus duplex, septate vagina, and hypoplasia of the uterus, ovaries, and fallopian tubes. Developmental delay, especially of fine and gross motor skills. A wide variety of socialization and social interaction problems have been identified with BBS. Obesity, possibly related to a decreased sensory function that would normally indicate satiation. Hyperphagia in some patients.[10]

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