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Case Report

Bardet- Biedl Syndrome: Two Case Reports

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Abstract

Bardet Biedl syndrome is a rare heterogeneous autosomal recessive disorder. A very few case was reported in Bangladesh. Here we two cases will be discuss .One is a 18 years young boy presented with childhood obesity (>97%), polydactyly in all 4 limbs, bilateral gynaecomastia, acanthosis nigricans, night blindness, mental retardation. After evaluation he was found to have hypogonadotrophic hypogonadism, primary hypothyroidism, dislipidaemia, renal impairment, elevated liver enzymes and retinitis pigmentosa. We advised him to reduce weight and implemented with weight reducing diet. Levothyroxine and Metformin were started. And another case is a 38 years unmarried lady presented with overweight, polydactyly in lower limbs, blindness, paraparesis and mental retardation with known Diabetes mellitus and hypertension. After evaluation she was found to normal hormonal activity and retinitis pigmentosa. She was also treated with anti- diabetic (Insulin) and antihypertensive medication. Both cases were scheduled for check-up every 2-3 months both in endocrinology and eye OPD.

Key Words: Bardet Biedl Syndrome, Retinitis Pigmentosa, Lawrence Moon Syndrome, Obesity, Hypogonadism.

Introduction:

Bardet Biedl syndrome is a genetically heterogeneous disorder that is usually inherited as autosomal recessive trait. The main problem is in the BBS1 gene located in the long arm of Chromosome 11.[1] Visual loss and obesity is the main feature of this syndrome. Other features are polydactyle or syndactyle, hypogonadism, impairment of speech, delayed motor skill development, distinctive facial appearance, behavioral abnormality, poor coordination, dental abnormalities and partial or complete loss of sense of smell. Here we have presented two cases of Bardet -Biedl syndrome with characteristics features associated with Metabolic Syndrome, Renal impairment, IGT, DM, Elevated liver enzymes and Hypertension.[2]

Case -1:

Mr. X, 18 years young boy, nondiabetic, normotensive, hailing from Manikgonj, Dhaka without any H/O consanguinity of marriage between parents, was admitted in BIRDEM General Hospital on 17th Aug 2017 with the complaints of night blindness, and under developed penis and scrotum. He is obese since childhood. There is learning difficulties with low IQ level. On query his mother complaints about increased hunger. His BMI is 31.98 Kg/m^2, which is more than 95 percentile according to his age. SPL is 2 cm and testicular volume is 1.5 ml for each. Tanner stage 1. Skin is thick, coarse and dry with presence of acanthosis nigricans. He also noticed Gynaecomastia (stage 4) on both side (Fig. 1). He has extra axial polydactyly in all 4 limbs (Fig. 2). Neurological examination of lower limbs reveals normal. Fundoscopy reveals retinitis pigmentosa (Fig. 3).

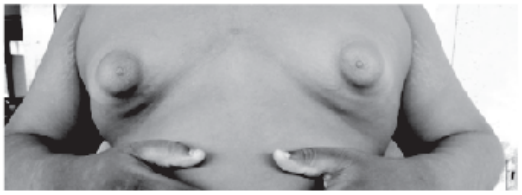

Fig. 1 Showing Gynaecomastia

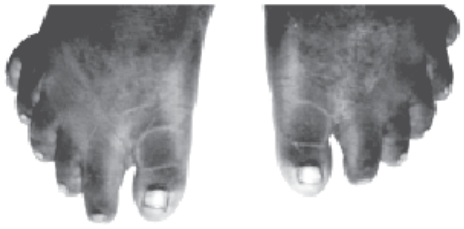

Fig. 2 Showing Polydactyly

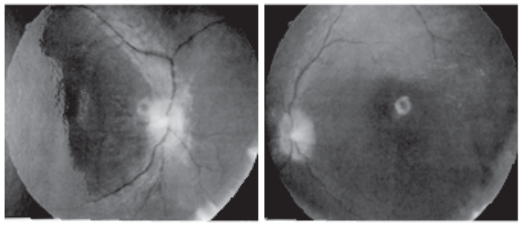

Fig. 3 Showing Retinitis pigmentosa

His hormone analysis reveals primary hypothyroidism (TSH- 8.09 ulU/ml, FT4- 14.64 pmol/L), Secondary hypogonadism (S. LH- 0.08 mIU/ml, S. testosterone- 0.36 ng/ml), USG W/A reveals fatty change in Liver. Fasting Lipid Profile reveals high Triglyceride level (176 mg/dl) decreased HDL level (35 mg/dl), And OGTT reveals IGT (Fasting 4.6 mmol/L and 2 hr after 75 gm glucose 8.4 mmol/L). His liver enzymes were also elevated including alkaline phospahtase (AST-141 u/L, ALT- 101 U/L, S. Bilirubin Level is 0.6 mg/dl and Alkaline phosphatase 318 u /L), His renal function tests shows mild renal impairment (S. creatinine- 1.3 mg/dl, eGFR-82.98 ml/min/1.73m2).

We advised the patient to do exercise regularly and implemented a weight reducing diet chart. We gave him levothyroxine 50 microgram once daily and metformin 500 mg twice daily. We requested him to come for follow-up after 1 and half month with the following reports: OGTT and S.TSH, FT4, Liver function tests, S.Creatinine. Ophthalmological follow up was scheduled after 3 months. In every follow up his BP will be monitored and sequential gonadal assessment will be done. If Puberty doesn't appear after 14 years of age, then plan for starting testosterone.

Case 2 :

Mis Y 38 years unmarried lady, diabetic, hypertensive coming from Dhaka was admitted in BIRDEM General Hospital with the complains of uncontrolled blood sugar .She has no H/O consanguineous marriage of her parents. On query her mother gave H/O gradual weight gain, diminished vision and subsequently developed blindness. She also complains of learning difficulties with low IQ level. Her BMI is 31.5 Kg / m2. There is an extra axial polydactyly (Fig. 4) on her lower limbs. Neurological examination of lower limb reveals diminished power (4/5) both proximally and distally. Fundoscopy shows non-proliferative diabetic retinopathy with retinitis pigmentosa.

No significant hormonal abnormality was detected. Her USG of W/A shows bright and coarse hepatic parenchyma with renal parenchymal disease. Liver

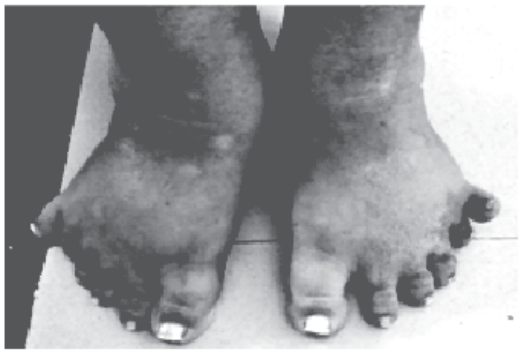

Fig. 4 Showing polydactyly

enzymes were also elevated (Alkaline phosphatase : 119 U/L and SGPT : 16 U/L) with grossly impaired renal function(Urea: 96 mg/dl, Creatinine: 4.2 mg/dl). Her HbA1c (8.9%) shows uncontrolled blood sugar).

We treated the patient with split mix human insulin for blood sugar control and advised to follow her diet chart. We also gave her Cilnidipine(10 mg) , Bisoprolol (5 mg) and Prazosine (5 mg) for blood pressure control. We also restricted her water and protein intake and requested her on regular follow up at endocrine, eye and nephrology OPD.

Discussion:

This syndrome is named after George Bardet and Arthur Biedl. The first known case was reported by Laurence and Moon in 1866 at the ophthalmology Hospital in south London. Laurence-Moon-Bardet-Biedl syndrome is no longer considered a valid term because Laurence and Moon had Paraplegia but no obesity or polydactyle, which is the key features of Bardet-Biedl syndrome. Laurence–Moon syndrome is now considered a separate entity.[3] Bardet-Biedl syndrome is rare, eg. occuring about 1 in 160000 population in Switzerland but It is more common in specific population, like 1 in 13500 among kuwaiti Bedouins and 1 in 17500 among residents of Newfoundland in Canada. Retinitis Pigmentosa is caused by progressive degeneration of Rod photo receptor cells in the retina. Prognosis is poor if there is renal involvement. Many patient die from renal failure in early age. Genetic counseling and pre-conception genotyping of family members may be worthwhile. Prenatal second-trimester ultrasound scanning in pregnancies with increased risk looking for polydactyly and renal anomalies (enlarged hyperechoic kidneys) can help in diagnosing BBS antenatally. There is no definitive treatment for BBS. Symptomatic treatment is the approach, like management of Obesity,

Cosmetic surgery for gynaecomastia, Using stick and support of caretakers for walking, multi-disciplinary approach from Speech therapist, Audiologist, Orthopedic surgeons and Endocrinologist.[4,5]

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