Bardet-Biedl Syndrome with Familial Retinitis Pigmentosa – A Case Report

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INTRODUCTION

Moon along with Laurence in 1866 documented a family of four siblings with obesity, spastic paraparesis, retinal dystrophy, and cognitive deficit.[1] Biedl[2] and Bardet[3] separately described later on people with similar features along with post-axial polydactyl and the syndrome was termed Laurence-Moon-Bardet-Biedl syndrome.[4] Five cardinal features of Bardet-Biedl syndrome (BBS) include polydactyly, pigmentary retinopathy, obesity, mental retardation and hypogonadism. Its prevalence is 1:13,500 in Kuwait.[5] Larger incidence in Arabic communities can be due to high rate of consanguineous marriages.[6] Less than 15 cases have been reported from India.[7]

Bardet-Biedl syndrome is a disorder of immotile cilia. Motile cilia generate movement of fluid and immotile cilia are sensory organelles defects which cause polydactyly, learning difficulties, retinitis pigmentosa, situs inversus, and cyst formation in pancreas, kidneys and liver.

The Bardet-Biedl syndrome is an infrequent autosomal recessive ciliopathic, pleiotropic disorder. It includes retinitis pigmentosa, obesity, polydactyly, learning disabilities and hypogonadism. This is a case series of a family with five out of eight siblings showing all ocular features and most of general features of Bardet-Biedl syndrome fulfilling the diagnostic criteria.

Diagnostic Criteria of the syndrome is enlisted below (Table 1). Presence of 4 major signs or 2 minor plus 3 major signs diagnoses this syndrome.

Major Features	Minor Features
	Learning difficulties
Polydactyly	Diabetes mellitus
Retinal dystrophy	Development delay
Central obesity	Facial dysmorphism
Renal abnormalities	Neurological deficits
Hypogenitalism in men	Hypertension
	Dental anomalies

Table 1. Diagnostic Criteria for BBS (Major and Minor Features)

PRESENTATION OF CASE

We report a series of five cases, five out of eight siblings fulfilling the diagnostic criteria for Bardet-Biedl syndrome. They were off springs of a second-degree consanguineous marriage.

Case 1

A 31-year-old man presented with a complaint of loss of vision in both eyes since childhood. He had diminution of vision in both eyes which progressed to blindness by the age of 8 years. He was the first among the eight siblings. He was mentally challenged. General examination showed polydactyl in all the limbs. The patient had a body-mass index (BMI) of 31.1. He had hypogonadism. He suffered from chronic kidney disease and was on haemodialysis. (Figure 1)

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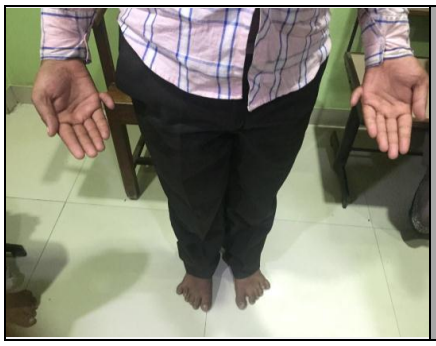

Figure 1. OCT of These Patients Typically Revealed Gross Foveal Thinning, with Absence of IS-OS Junction and Photoreceptor Layer with Enhanced Choroidal Visualisation

Both eyes had no PL (Perception of Light). Intraocular pressures (IOP) was 24 and 25 mm of Hg in right and left eye respectively. He had posterior subcapsular cataract in both eyes. On fundus examination a waxy pale disc, attenuation of arteries, tessellated background and bony spicules in the periphery, were seen suggestive of retinitis pigmentosa with optic atrophy bilaterally.

Case 2

26-year-old woman came complaining of blurring of vision at night since childhood which has slowly progressed to diminished vision even during the daytime. She is the third among the 8 siblings. She was mentally challenged. General examination showed polydactyl in both upper limbs. She had a BMI of 33.1. At presentation her vision was hand movements close to face in both her eyes. IOP measured 22 mm of Hg bilaterally. Anterior segment findings on slit lamp examination were normal. Fundi of both eyes showed typical features of retinitis pigments with characteristic optical coherence tomography (OCT) pattern of foveal thinning, with absence of inner and outer segment junction (IS-OS) with enhanced choroidal visualisation.

Case 3

22-year-old man came complaining of blurring of vision in both eyes since childhood especially in the night, which progressed gradually over the years. He is the 4th among the 8 siblings. General examination showed polydactyly in upper 2 and right lower limb. Patient had a BMI of 29.1. He had hypogonadism. He is a diagnosed case of chronic renal disease under treatment. Visual acuity was hand movements near to face bilaterally. His IOP taken was 18 and 20 mm of Hg in right eye and left eye respectively. Anterior segment had early posterior subcapsular cataract in both eyes. Fundi of both eyes showed a waxy pale optic disc and pigmentation in the retinal periphery consistent with retinitis pigmentosa.

Case 4

An 18-year-old woman came with diminution of vision bilaterally. She's is the 6th among the 8 siblings. She was mentally challenged. General examination revealed polydactyly in left upper limb. Her BMI was 33.6. Her vision was hand movements close to face in her right eye and finger counting at 1 metre in her left eye. IOP was 18 and 19 mm of Hg in right and left respectively. Case presented with posterior subcapsular cataract bilaterally and fundi of both eyes showed findings suggestive of retinitis pigmentosa.

Case 5

A 14-year-old boy, 8th offspring of the family, presented with complains of diminution of vision in both eyes since childhood which is gradual and progressive in nature. The boy was mentally challenged. General examination revealed polydactyly in upper limbs and BMI -29.4. His vision was counting fingers and hand movements in both eyes. Intraocular tension was 12 mm of Hg in right and 11 mm Hg in contralateral eye. Anterior segment on slit lamp was normal. Fundus documented retinitis pigmentosa in both eyes.

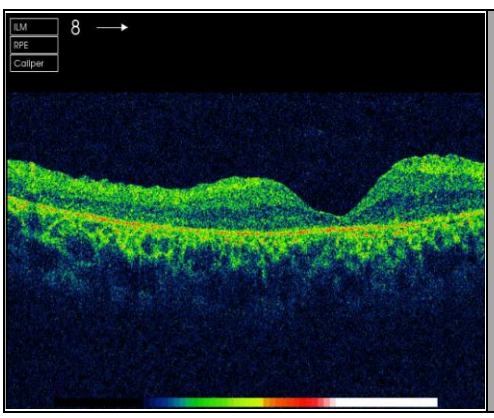

Figure 2. Overview of Clinical Features

Case No	Age	Gender	Obesity	Retinitis Pigmentosa	Mental Retardation	Polydactyly	Hypogonadism	Renal Abnormality
1	31	Male	+	+	+	+	+	+
2	26	Female	+	+	+	+	x	x
3	22	Male	+	+	x	+	+	+
4	18	Female	+	+	+	+	x	x
5	14	Male	+	+	+	+	+	x-

Table 2. All Five Patients were Diagnosed with Biedl-Bardet Syndrome Based on the Diagnostic Criteria

DISCUSSION

Bardet-Biedl syndrome is a pleiotropic inherited disorder with notable interfamilial and intrafamilial variation.[8,9] The disease does not have any known modifiable or preventive risk factors except avoidance of consanguineous marriage.

Apart from the cardinal manifestations, other features include congenital heart block, brachycephaly, dental anomalies and deafness. Early and prompt recognition is important to help in the treatment of the child through timely assessment of blood pressure, weight, renal imaging and renal function test, ophthalmologic examinations and psychological support although no definitive treatment at present.[10]

Rapid neonatal diagnosis achievable by correlating prenatal detection of polydactyly with sonography along with genetic studies shortly after birth[11] Nephrologists have a main role in management as renal failure most likely causes death

among Bardet-Biedl syndrome patients.[12] Testosterone supplements may be given to male sufferers, specifically in cases having lowered level of this hormone.[13]

Accessory fingers and toes are often non-functional and can be excised. Obesity is a major worrisome area as, if left uncontrolled, will lead to multiple health problems. A low-protein and low-calorie diet helps to combat obesity and slows the progression of renal failure.

There are no documented effective treatments to either avoid or reduce the deterioration in vision. However, low vision aids can be helpful to the patient, and regular ophthalmological follow-up is stressed upon.

The detrimental effects of consanguineous marriage cannot be overlooked. In many communities, in whom the frequency of consanguinity in marriages is large, there is an associated low genetic literacy level among the public so the need for education is very important for them.

The offspring of consanguineous unions may have a high chance to present with recessive disorders due to the appearance of recessive genetic mutations received from a common ancestor. The nearer a genetic association of parents, more is the chance their child will inherit similar copies of detrimental recessive genes.[14]

CONCLUSIONS

In such societies, wherein in the rate of consanguineous marriages is very large are mostly poor in genetic knowledge among the general public including the health care personnel. So, the requirement for education is of utmost need. Premarital and preconception genetic counselling is essential in populations wherein consanguineous marriage is still practiced and abortion of the affected fetus is not accepted by the community. This is the largest number of cases reported of Bardet-Biedl syndrome in a family.

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