INTERNATIONAL
AYURVEDIC
MEDICAL JOURNAL

IAMJ

Case Report ISSN: 2320 5091 Impact Factor: 4.018

BARDET-BIEDL SYNDROME – A RARE GENETIC DISORDER

Shine S. Nair

Assistant Professor, Department Of Kaumarabhritya, Rajiv Gandhi Ayurveda Medical College, (Govt. of Puducherry Institution), Mahe, Puducherry, India

Email: drshinesnair@gmail.com

ABSTRACT

Bardet – Biedl syndrome is a cliopathic human genetic disorder that produces many effects and affects multiple systems. It is characterised by obesity, retinitis pigmentosa, polidactyl, hypogonadism and in some cases kidney failure. Post-axial polydactyly is common and may be the only obvious dysmorphic feature at birth. Diagnosis is based on clinical features. The most common diagnostic handle prompting investigation for BBS is the development of rod-cone dystrophy. Primary loss of rod photoreceptors is followed by later demise of cone photoreceptors. This presents as an atypical retinitis pigmentosa. Obesity is another major clinical finding and the incidence is reported to be 72–86% in the BBS population. Hypogonadism may manifest as delayed puberty or hypogenitalism in males and genital abnormalities in females. Developmental delay and cognitive deficit are common in BBS. Speech deficit has been reported in 60% of patients. Speech difficulties may be complicated by hearing loss, which is reported in 17–21% of patients.

Keywords: Retinitis pigmentosa, Polydactyly, Hypogonadism

INTRODUCTION

In 1866, Laurence and Moon described a family of four siblings with retinal dystrophy, obesity, spastic paraparesis and cognitive deficit. Bardet and Biedl later reported separately on further similarly affected individuals who in addition had post-axial polydactyly and the condition was coined Laurence–Moon–Bardet–Biedl syndrome. The syndrome is often divided into two entities: Laurence–Moon syndrome and Bardet–Biedl Syndrome (BBS), but there is considerable phenotypic overlap. BBS is now the standard term in common usage.

Bardet–Biedl syndrome is a rare autosomal recessive ciliopathy characterised by retinal dystrophy, obesity, post-axial polydactyly, renal dysfunction, learning difficulties and hypogonadism. The diagnosis is based on clinical findings. The BBS phenotype evolves slowly throughout the first decade of life, although there is a considerable variability. As a result, most patients are diagnosed in late childhood or early adulthood. Post-axial polydactyly is common and may be the only obvious dysmorphic feature at birth.

The most common diagnostic handle prompting investigation for BBS is the development of rod-cone dystrophy. Primary loss of rod photoreceptors is followed by later demise of cone photoreceptors. This

presents as an atypical retinitis pigmentosa with early macular involvement. The clinical manifestation is gradual onset of night blindness, followed by photophobia and loss of central and colour vision

Obesity is another major clinical finding and the incidence is reported to be 72–86% in the BBS population. Birth weight is usually within the normal range. One-third of those with a normal birth weight develop obesity by the age of one. Although adult obesity tends to be truncal, it appears to be widespread and diffuse in childhood. The development of type 2 diabetes is prevalent among patients.

Hypogonadism may manifest as delayed puberty or hypogenitalism in males and genital abnormalities in females. This may occur independently or in conjunction with biochemical hypogonadism. A wide range of genital malformations have been observed in females, contributing to the low rates of fertility in BBS. Males are almost always infertile.

Developmental delay and cognitive deficit are common in BBS. Delay is often global but may be specific to certain areas of development. Renal abnormalities can be a major cause of morbidity and mortality in BBS. The renal phenotype is variable but classically manifests with cystic tubular disease and anatomical malformations.

Speech deficit has been reported in 60% of patients. This mainly consists of high-pitched nasal speech and children often do not develop understandable speech before the age of four. Speech difficulties may be complicated by hearing loss, which is reported in 17–21% of patients. Most patients suffer conductive hearing loss secondary to chronic otitis media. Speech delay in children with BBS is generally responsive to speech therapy.

Modified diagnostic criteria by Beales suggest that either four primary features or three primary and two secondary features are required to make a clinical diagnosis.

Table 1:

Primary features	Secondary features
Rod-cone dystrophy	Speech delay
Polydactyly	Developmental delay
Obesity	Diabetes mellitus
Genital anomalies	Dental anomalies,
Renal anomalies	Brachydactyly/ Syndactyly
Learning difficulties	

MANAGEMENT

A multidisciplinary approach is required to effectively manage this pleiotropic condition. Although research is in progress, there is still no targeted treatment for BBS. Complications associated with BBS should be treated symptomatically as in the general population.

CASE REPORT

History of present symptoms:

A 9 Year old male child was brought to the outpatient department with complaints of distension of abdomen (obesity), blurred vision, lack of interest in study and reduced memory power.

Past history:

He was the first sibling of consanguineous marriage. He was delivered at full term by normal vaginal delivery. Baby cried immediately after delivery. The birth weight was 3.5kg.There was no significant post natal events like pathological jaundice, sepsis etc. His developmental milestones were slight delayed. He had been completely immunized.

On Examination:

The child was obese. He was moderate built. His anthropometric measurements include, the height was 129cm, the weight was 35kg, the head circumference 49cm, the abdominal girth was 66cm and the chest circumference was 70cm.He was afebrile. Pulse rate was 70 per min, the respiratory rate was 30 per min. the blood pressure recorded was 120/80mm of Hg. He

was having small and undeveloped sexual characters (undeveloped testis and penis), difficulty in identifying and naming objects. There was no pallor, no lymphedinopathy, no cyanosis and no clubbing. Other systemic examinations were normal.

***Case at a glance**:*

Height: 129 cm, Pulse: 70 /min, Respiratory rate: 30 /min, Blood pressure: 120/ 80 mm of Hg

***Positive findings**:*

- History: Consanguineous marriage, Obesity, Lack of interest in studies, Delayed milestones, Short structure, Blurred vision
- Examination: Small and undeveloped sexual characters (micro penis and testis), Increased abdominal girth, Reduced head circumference, Polydactyly (extra fingers and toe), Speech difficulty

Investigations done:

Blood Examination**:** Hb: 12gm%, TC: 10250cells/cmm, ESR: 16mm/hr, DC: N- 60%; L- 35%; M- 1%; E- 4%, RBS: 88.2 mg/dl, Blood urea: 15.1 mg/dl

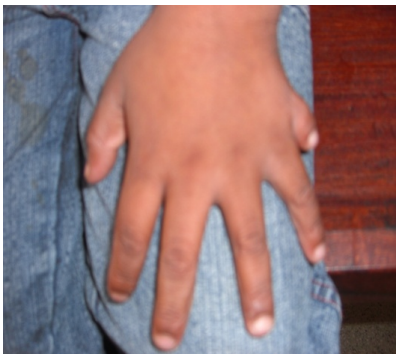
Urine examination: Albumin: Absent, Sugar: Absent, Pus cells: 2-3, Epithelial cells: 2-4

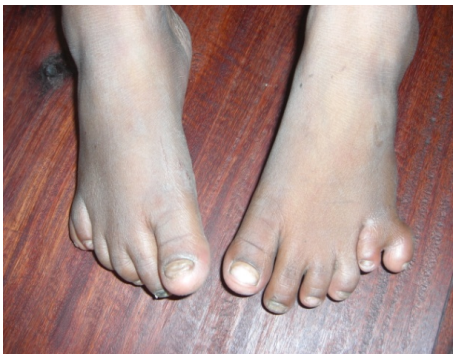
Thyroid function test: T3 – 170 mg/dl, T4 – 10.40 mg/dl, TSH – 3.22Micro IU/ml

Differential diagnosis:

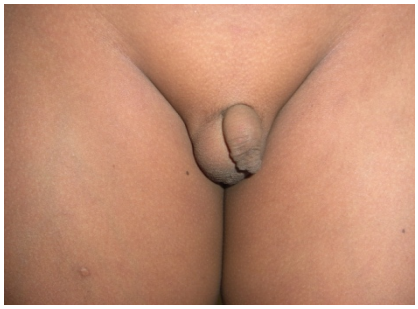
Obesity, Hypothyroidism, Cushing's syndrome, Prader-Willi syndrome, Frohlich's syndrome, Laurence- Moon syndrome, Bardet-Biedl syndrome

PHOTOGRAPHS

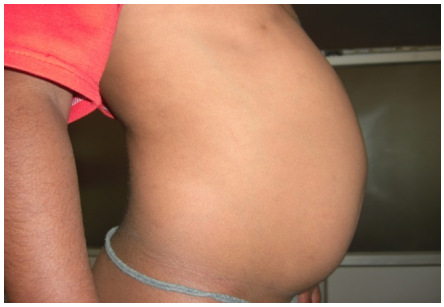




Polydactyly (Extra fingers and toe)



Small and undeveloped sexual characters



Obesity

CONCLUSION

Based on the positive findings like consanguineous marriage, obesity, lack of interest in studies, delayed milestones, short structure, blurred vision, small and undeveloped sexual characters, increased abdominal girth, reduced head circumference, polydactyly and speech difficulties the case was diagnosed as **BARDET-BIEDL SYNDROME.**

REFERENCES

1. Laurence JZ, Moon RC: Four cases of 'retinitis pigmentosa' occurring in the same family, and accompa-

nied by general imperfections of development. *Obes Res* 1995; 3: 400–403.
2. Bardet G: On congenital obesity syndrome with polydactyly and retinitis pigmentosa (a contribution to the study of clinical forms of hypophyseal obesity). *Obes Res* 1995; 3: 387–399.
3. Biedl A: A pair of siblings with adiposo-genital dystrophy. *Obes Res* 1995; 3: 404.
4. Beales PL, Elcioglu N, Woolf AS, Parker D, Flinter FA: New criteria for improved diagnosis of Bardet–Biedl syndrome: results of a population survey. *J Med Genet* 1999; 36: 437–446.
5. Hamel CP: Cone rod dystrophies. *Orphanet J Rare Dis* 2007; 2: 7.
6. Baker K, Beales PL: Making sense of cilia in disease: the human ciliopathies. *Am J Med Genet C Semin Med Genet* 2009; 151C: 281–295.
7. Putoux A, Attie-Bitach T, Martinovic J, Gubler MC: Phenotypic variability of Bardet-Biedl syndrome: focusing on the kidney. *Pediatr Nephrol* 2012; 27: 7–15.
8. O'Dea D, Parfrey PS, Harnett JD, Hefferton D, Cramer BC, Green J: The importance of renal impairment in the natural history of Bardet-Biedl syndrome. *Am J Kidney Dis* 1996; 27: 776–783.
9. Beales PL, Warner AM, Hitman GA, Thakker R, Flinter FA: Bardet-Biedl syndrome: a molecular and phenotypic study of 18 families. *J Med Genet* 1997; 34: 92–98.

Source of Support: Nil
Conflict Of Interest: None Declared

How to cite this URL: Shine S Nair: Bardet-Biedl Syndrome – A Rare Genetic Disorder. International Ayurvedic Medical Journal {online} 2019 {cited March, 2019} Available from: http://www.iamj.in/posts/images/upload/492_495.pdf