Case Report

University J Dent Scie 2021; Vol. 7, Issue 2

Oral-Systematic Manifestation of Bardet- Biedl Syndrome: A Rare Case Report.

Abstract :
Bardet-Biedl Syndrome (BBS) is a rare, heterogenous, ciliopathic autosomal recessive condition, affecting multiple organs. Primary features includepolydactyly, obesity, mental retardation, hypogonadism, retinopathy and renal failure. Dental anomalies is regarded assecondary features, include hypodontia, microdontia, short roots and deep palate. Less than 15cases have been reported from India. This article reports a case of Bardet-Biedl Syndrome in a male with typical systemic and oral features. This case exemplifies the need of multidisciplinary approach.

Keywords : Bardet- Biedl Syndrome, Dental anamolies

Introduction:

Bardet-Biedl syndrome (BBS) is a rare pleiotropic genetic disorder with severe multi organ impairment.1 The frequency of the syndrome is estimated to be [1]. 1,60,000.[2] Very few cases have been reported from India. 3The incidence is much higher in populations with a high level of consanguineous marriage or areas those are geographically isolated. For example disease incidence is 1 in 13,000 and 1 in 170000 live births in the isolated populations of Newfoundland and Kuwait respectively[4], where high incidence of marriage between relative persists.

BBS is genetically heterogeneous with at least 6 loci mapped to date. BBS genes encode proteins that localize to the cilia and basal body and are involved in cilia biogenesis and function[5] The most common gene is BBS1 located at chromosome 11.[6] Many individuals with BBS look virtually healthy at birth unless if polydactyly is present at birth. Patients diagnosed with BBS at early child hood have fewer clinical features. Usually other features slowly emerge with aging. The primary and secondary features of BBS are described in Table 1. The disease symptoms may significantly vary from patient to patient therefore, the diagnosis relies on the number of primary and secondary features of BBS.

Access this article online

Website: www.ujds.in

DOI: https://doi.org/10.21276//ujds.2021.7.2.18

Quick Response Code

Diagnosis of BBS requires the presence of at least four primary or three primary and two secondary clinical features.[7, 8]

Dental anomalies are considered as secondary features of BBS which include high arched palate, small teeth, hypodontia and small teeth.

There are only few reports describing oral and systemic manifestation of this syndrome. Here we are reporting a rare classical case of BBS having oral and systematic manifestation.

Case Description:

A 27 years patient suffering from BBS was referred to Department of Periodontics RUHS-College of Dental Science,Jaipur from Department of Medicine SMS hospital

[1]DEBOPRIYA CHATTERJEE, [2]ANJALI KAPOOR
[3]SANJEEV KUMAR, [4]MEENAKSHI SHARMA
[1,2]Department of Periodontics, RUHS- College of Dental Sciences, Jaipur
[3]Department of Cardiology, SMS Medical College and Hospital
[4]Department of Pedodontics, RUHS- College of Dental Sciences, Jaipur

Address for Correspondence: Dr. Debopriya Chatterjee MDS, Senior Demonstrator, Department of Periodontics, RUHS-College of Dental Sciences, Jaipur
Email : banerjee.debo@gmail.com

Received : 10 June, 2021, **Published :** 31 August 2021

How to cite this article: Chatterjee, D. D., Kapoor, D. A., Choudhary, D. S., & Sharma, D. M. (2021). Oral - Systematic manifestation of Bardet- Biedl Syndrome: A rare case report. UNIVERSITY JOURNAL OF DENTAL SCIENCES, 7(2).: 88-90

Jaipur for dental opinion. Patient had poor oral hygiene with halitosis. Patient gave a history of bleeding from gums. Based on his clinical features, patient was diagnosed as having BBS at the age of 11 years.

The patient gave a history of consanguineous marriage among his parents. There was a history of death of one his younger female sibling and one younger male sibling who is also suffering from BBS. On physical examination patient has diminished vision especially at night. Icterus was present in eye and fundus examination was suggestive of retinitis pigmentosa [Fig 1]. Patient was mentally retarded with I. Q level less than 70. The patient body mass index was 40, which indicates severe obesity. He had short and stubby limbs. Truncal obesity was present with distention of stomach. There was post axial polydactyly with hexadactyly of all four limbs [fig2, 3].

Liver function tests were deranged, SGOT/PT was 130/75, PT/INR 19.8/1.72, total serum bilirubin was 7.9. Routine urine microscopy findings were normal. Blood and urine culture sensitivity were negative. Viral markers (Anti Hcv, Hbs Ag, Anti HAV Ig M, Anti H E V Ig M and HIV) were negative. Ultra sound and 2D ECHO revealed of splenomegaly and global hyperkinesias of left ventricle respectively. Both kidneys were normal in functions. Left adrenal glands is enlarged. Patient had moderate to severe hearing loss.

An extra oral examination showed retrognathia, hypotonic upper lips, forceful closing of lips and mouth breathing was present [fig 4]. An intra oral examination revealed high arched palate with anterior deep bite [Fig 5].There was bilateral loss of mandibular first molar. No abnormality was detected in tongue. There was facial asymmetry, with drooping of right corner of mouth. Facial nerve was normal on examination. There was severe bacterial plaque index, presence of carious lesion, slight flourosis. Generalized clinical attachment and bone loss was present. Grade I mobility was present on all teeth. Patient was provided with oral prophylaxis. Instructions were given to mother to brush her son's teeth. Patient was instructed to rinse with 0.12% chlorhexidine gluconate twice a day for 1 week. No evidence of skeletal dysplasia and abnormality of the spinal cord or shoulder were detected. The patient had learning disabilities and was lagging to follow verbal commands. The parents gave history of his inability to stand by himself at the age of 5 years.

A multi disciplinary approach is required to manage this pleiotropic condition.

Discussion:

Bardet-Biedl Syndrome is an uncommon, Autosomal heterogenous ,recessive disorder. The diagnosis of BBS is based on clinical criteria.9Bardet-Biedl syndrome is named after Georges Bardet and Arthur Bied.[10] The syndrome is due to defect in BBS gene, encoding BBD protein . This protein, in particular related to ciliary and centrosomal activities as it assist in microtubule-related transport and cellular organization processes.[11] Some of the phenotypes exhibited due to BBS protein sabnormility, including retinal degeneration, skeletal anomalies and renal cysts/malformations are similar to human diseases associated with ciliary disfunction.[12] The treatment of Bardet-Biedl syndrome is symptom and subject specific that are apparent in each individual. Multi disciplinary approach of Pediatricians, orthopedic surgeons, cardiologists, dental specialists, speech pathologists, audiologists, ophthalmologists, nephrologists, and other healthcare professionals is required for treatment. Individuals with Bardet-Biedl syndrome should undergo regular periodic examinations. Blood pressure should be measured once in six months, as there is chances of development of hypertension.[5] Periodic detailed opthalmological assestment is required. Patient may also require necessary review from clinical psychologist to deal any kind of behavioral issues like anxiety, depression. As well as periodic systemic assessment of disorder such as kidney dysfunction, diabetes mellitus, liver function as they are frequent in BBS patients. Depending on the need patient may require a regular visit to periodontist for assessment of periodontal problems and orthodontist for assessment of dental crowding / hypodontia.[13]

Table 1

Primary and Secondary Features of Bardet-Biedl Syndrome	
Primary Features	Secondary Features
• Retinal dystrophy • Polydactyly • Obesity • Developmental delay • Hypogonadism in males • Renal anomalies	• Speech disorder/delay • Strabismus/cataracts/astigmatism • Brachydactyly/syndactyly • Polyuria/polydipsia (nephrogenic diabetes insipidus) • Diabetes mellitus • Ataxia/poor coordination/imbalance • Mild spasticity (especially lower limbs) • Hypodontia/smallroots/high archedpalate/small teeth • Left ventricular hypertrophy/congenitalheart disease Hepatic fibrosis

Conclusion:

Bardet-Biedl syndrome is a very rare, genetic syndrome. The syndrome has various phenotype expressivities. Diagnosis is based on criteria with 4 primary or 2 primary and 2secondary features. Early diagnosis and intervention is important in ensuring that children with BBS have improved quality of life. Genetic counseling may also benefit the affected individual and their family. Treatment is directed towards symptom specific and requires coordinated team of specialist.

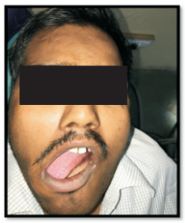

Figure1:- Frontal view

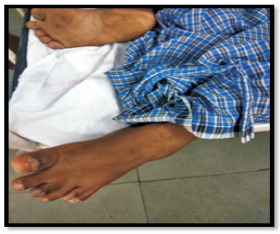

Figure 2:-post axial polydactyly of hands

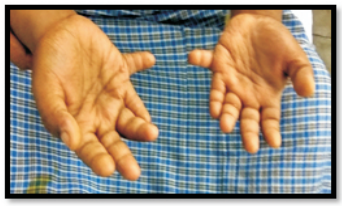

Figure 3:- post axial polydactyly of limb

Figure 4:-Facial profile showing hypotonic lips

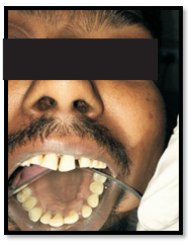

Figure 5:- intraoral palatal view

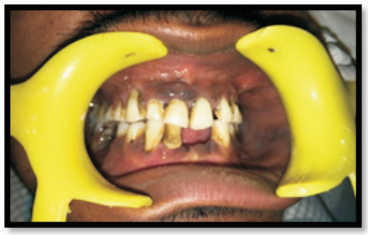

Figure 6:-intraoral labial view

References:

1. Waters AM, Beales PL: Bardet–Biedl Syndrome. Gene Reviews [Internet] 1993-2003 (Updated 2011).
2. Klein D, Ammann F. The syndrome of Laurence-Moon-BardetBiedl and allied diseases in Switzerland. Clinical, genetic and epidemiological studies. J NeurolSci 1969; 9:479-513.
3. Hooda AK, Karan SC, Bishnoi JS, Nandwani A, Sinha T. Renal transplant in a child with Bardet-Biedl syndrome: A rare cause of end-stage renal disease. Indian J Nephrol 2009; 19:112-114.
4. Moore SJ, Green JS, Fan Y, Bhogal AK, Dicks E, Fernandez BA, et al. Clinical and genetic epidemiology of Bardet-Biedl syndrome in Newfoundland: A 22-year prospective, population based, cohort study. Am J Med Genet 2005; 132:352-6.
5. KristineT.Lo. Manifestations of Bardet-Biedl Syndrome. Philippine Journal of Opthalmology 2004; 29:94-98.
6. Mykytyn K, Nishimura DY, Searby CC, Shastri M., et al Identification of the gene (BBS1) most commonly involved in Bardet-Biedl syndrome, a complex human obesity syndrome. Nat Genet 2002; 31: 435-438.
7. 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.
8. Forsythe E, Beales PL: Bardet-Biedl syndrome. Eur J Hum Genet 2013;21:8–13.
9. Sahu JK, Jain V. Laurence- Moon –Bardet-Beidl – Syndrome. J Nepal Med Assoc 2008;47(172):235-237.
10. KharcheJyoti et al. Bardet-Biedl Syndrome- A Rare Case PresenatationInMarathwada Region Of India.IOSR-JDMS 2017; 16(7): 49-52.
11. Ansley SJ, Badano JL, Blacque OE, Hill J, Hoskins BE, Leitch CC, et al. Basal body dysfunction is a likely cause of pleiotropic Bardet-Biedl syndrome. Nature 2003; 425:628-633.
12. Pazour GJ, Rosenbaum JL. Intraflagellar transport and cilia-dependent diseases. Trends Cell Biol 2000;12:551–555.
13. Genuis SJ, Lobo RA: Potential amelioration of morbidity in patients with chromosomal anomalies: relevance to Bardet-Biedl syndrome. Clin Genet2011; 79:482–488 .