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Bardet-Biedl Syndrome: A Rare Case Reported in Two Siblings of Muzaffarnagar District, India

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Abstract: Bardet-Biedl syndrome (BBS) is a rare autosomal recessive genetic disorder belonging to the family of ciliopathies and characterized by heterogeneous clinical manifestations including rod-cone dystrophy, polydactyly, obesity, genital abnormalities, renal defects, learning difficulties, developmental delay, speech deficit, brachydactylic or syndactyly, dental defects, ataxia or poor coordination, olfactory deficit, diabetes mellitus, congenital heart disease, etc. Here, we present the case report of BBS, reported in two siblings, which is very rare.
Keywords: Bardet-Biedl syndrome, retinitis pigmentosa, hypogonadism.

INTRODUCTION

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive genetic disorder belonging to the family of ciliopathies. It is characterized by heterogeneous clinical manifestations including primary features of the disease (rod-cone dystrophy, polydactyly, obesity, genital abnormalities, renal defects, and learning difficulties) and secondary BBS characteristics (developmental delay, speech deficit, brachydactylic or syndactyly, dental defects, ataxia or poor coordination, olfactory deficit, diabetes mellitus, congenital heart disease, etc.); most of these symptoms may not be present at birth but appear and progressively worsen during the first and second decades of life [1].

It is transmitted by autosomal recessive inheritance, in which parents are heterozygotes and asymptomatic. Siblings have a 25% chance of being healthy, 50% will be heterozygous and 25% will be affected by the disease [2]. The prevalence BBS is 1 in 13500 to 160000 individuals depending on geographic location. The incidence is much higher in some populations with a high level of consanguinity [3]. Less than 15 cases have been reported from India [4]. Here, we present the case report of BBS, reported in two siblings.

CASE REPORT

A 15-year-old female patient was reported to us in the department of ophthalmology with complaints of night blindness, excessive weight gain, and menstrual irregularities. Secondary sexual characters are also not well developed in this patient. She was born to consanguineous marriage. She also has learning disabilities. There was the presence of postaxial polydactyly with hexadactyly of feet [Figure 1] and characteristic feature of acanthosis nigricans in this patient. Her height and weight were 134cm and 57kg respectively, with a BMI of 31.74 Kg/m^2, which indicated that the patient was obese. Her 7-year-old brother presented with complaints of behavioral issues and diminished vision, especially at night. He has got the characteristic feature of hypogonadism and micropenis (<2.5cm) [Figure 2]. There was the presence of postaxial polydactyly with hexadactyly of hands in a male sibling [Figure 3].

Hearing assessment was done which was found to be normal in both the patients. In order to rule out underlying cardiac abnormalities, we have done echocardiogram and electrocardiogram in both the cases and were found normal. Ultrasound examination revealed bilateral small ovaries in female patients and a renal anomaly in both male and female patients. The assessment of IQ was performed on both the patients. The IQs of both the patients were found to be less than 70, which is suggestive of mild mental disability. Fundus examination of both the patients shows bilateral retinitis pigmentosa.

Fig-1: Postaxial polydactyly with hexadactyly of feet of female patient

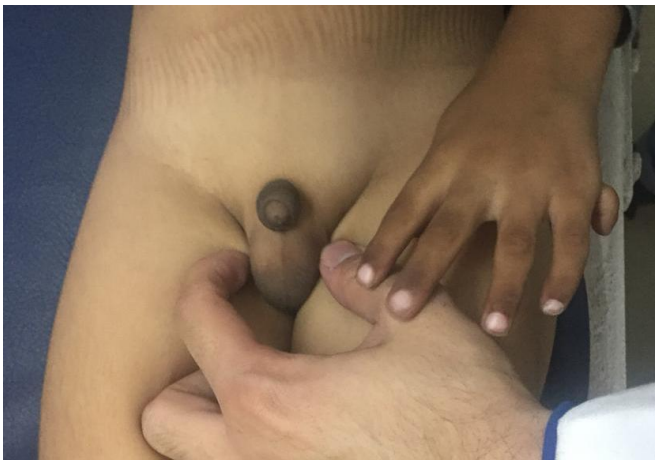

Fig-2: Micopenis in male patient

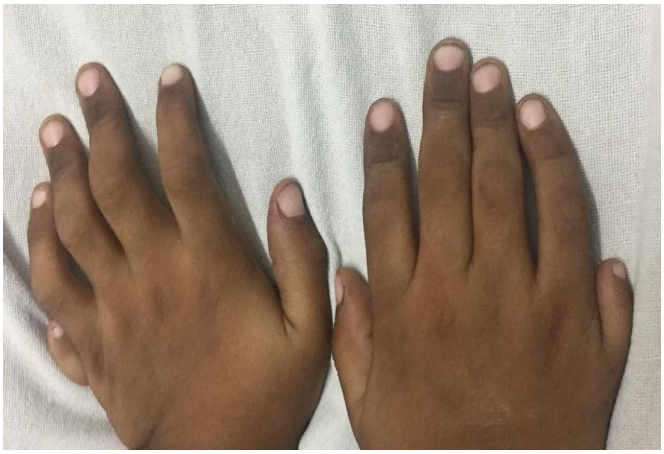

Fig-3: Postaxial polydactyly with hexadactyly of hands in a male sibling

We have done a complete blood count (CBC), which was found normal in both the patients. Besides normal CBC, biochemical analysis i.e. liver function tests (LFT), renal function tests (RFT) were found to be normal. Blood sugar analysis revealed slightly increased blood glucose in female patients, suggestive of prediabetes whereas in a male patient, the glucose level was within normal range.

DISCUSSION

BBS is named after Georges Bardet and Arthur Biedl. The first known case was reported by Laurence and Moon in the year1866. Laurence–Moon–Biedl–Bardet syndrome (LMBBS) is no longer considered as a valid term as patients of Laurence and Moon had paraplegia but no polydactyly and obesity, which are the key elements of the BBS. Hence, Laurence–Moon syndrome is typically taken into consideration a separate entity [5]. The exact biochemical mechanism that leads to BBS is not clear yet. Twelve genes (BBS1

to BBS12) that are responsible for the disease have been identified. The BBS proteins are components of the centrosome and affect the ciliary transport; hence, the disease falls under the spectrum of "ciliopathies" [6]. The most frequent finding of BBS is retinitis pigments, which we found in our both the patients. Retinopathy occurs in 90% of the patients of BBS. Retinal dysfunction usually becomes apparent at age 7–8 years, when night blindness gradually starts [7]. Obesity which is the second most common clinical findings of BBS is an area of major concern in these patients and if left uncontrolled, will lead to multiple health problems [8]. We also found both the patients were mentally retarded. Severe learning disabilities such as the inability to read or write have been found in BBS patients, making them completely dependent on parental care [9]. Hypogonadism is much more common in males compared to females. Hypogonadism in females results in failure of development of secondary sexual characteristics and delayed menarche. Males have a micropenis at birth, with small volume testes [10].

CONCLUSION

Our patients present with the classical features of BBS and this is a very rare case, which was reported in two siblings. So, we have presented the above cases of BBS. Though BBS is not curable, they were advised for regular follow up in order to monitor the problems associated with BBS.

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