

Siblings with Bardet Beidl Syndrome

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Introduction

Bardet Beidl syndrome (BBS) is an autosomal recessive condition characterized by central obesity, mental retardation, hypogonadism, renal dysfunction, post axial polydactyly, retinal dystrophy¹. This condition is more common in Newfoundland Island (1 in 17000) and Bedouin population of Kuwait (1:13500)². Because of the heterozygosity of BBS gene, there is a high prevalence that the unaffected family members can have renal dysfunction & renal cell carcinoma. There are four loci mapped till now (BBS1, BBS 2, BBS3, BBS4). BBS1 is the most common involves affected among white families & BBS4 is the next common^{3,4}. Diagnosis is quite difficult as the variants are very common within and between families. Clinical manifestations become evident in different ages like obesity by 2-3 years and retinal changes by a mean age of 8.5 years. Night blindness becomes evident by 7-8 years. There has been a criteria that proposed by Schachat and Maumenee involves retinal degeneration, mental retardation, obesity, polydactyly and hypothyroidism. Out of four by five if present, this syndrome can be diagnosed⁵. Here are with two siblings one with hypothyroidism without polydactyly and another with polydactyly.

Case 1

Elder sibling nine year old female who had global developmental delay presented with gaining weight inappropriately, decreased activity, decreased mingling with peer groups, speech delay, academically very poor performance, decreased vision especially at night for the last six years. There is also history of polyuria and polydypsia. No history of headache or abdominal pain. On examination child is short in stature (Height <3rd percentile) and obese (Weight >95th percentile). Her SMR is stage I. she has got depressed nasal bridge,

Abstract

Bardet Biedl syndrome is an autosomal recessive condition affecting many parts of the body. Incidence of BBS is 1 in 100000. Its clinical features varies in person to person though from same family too. We are reporting two siblings with Bardet Beidl syndrome with different clinical presentation.

Key words: Bardet Beidl Syndrome, Hypothyroidism, Retinal degeneration

widened philtrum, micrognathia, microphthalmia, vertical nystagmus, night blindness, retinal degeneration, epicanthal folds, short broad limbs, acanthosis nigricans, low hair line and hepatomegaly. The swinging mood is also observed in her.

Case 2

Younger one five year old male who also has global developmental delay presented with decreased mingling with peer groups, decreased activity and speech delay. He is also obese, moody, with depressed nasal bridge, widened philtrum, short neck, polydactyly, short broad limbs, short gaze, small penis (3 cm), small but descended testis, protrudent abdomen and saddle gap. His height was in <3rd percentile and weight was >95th percentile. Both of them had uneventful birth history and the family history is unremarkable.



Fig 1: Showing feet of sibling 1 with Polydactyly

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Fig 2: Showing hands of sibling 2 with Polydactyly.



Fig 3: Showing both the siblings having obesity and short stature.

Discussion

The clinical manifestation of BBS has been described initially by Bardet & Biedl in 1920.⁶ BBS is associated with obesity, limb abnormalities, hypogonadism, mental retardation, early onset of retinal degeneration and renal disease. The features that can be present along with this are hepatic fibrosis, diabetes mellitus, reproductive abnormalities, endocrinological disturbances, short stature, developmental delay and speech deficit, behavioral abnormalities, eye abnormalities, ataxia, orodental abnormalities and cardiovascular abnormalities, Hirschsprung's disease,

anosmia, craniofacial dysmorphism¹. Because of the heterozygosity of BBS gene, there is a high prevalence that the unaffected family members can have renal dysfunction & renal cell carcinoma. The common condition to be differentiated is Lawrence Moon syndrome, where mental retardation, hypogonadism, retinal degeneration occurs along with progressive neurological deficit, distal muscle weakness and there won't be any polydactyly. Retinal degeneration is the first major feature of this syndrome present in about 100% by second decade of their life⁷. The defect is described as an atypical pigmentary retinal dystrophy of photoreceptors with macular involvement⁸.

Obesity is the second major feature of BBS, present in about 72 – 96% cases. With age advances, the severity also increases. Limb abnormalities are the third specific feature of BBS. Limb deformities like polydactyly, post axial polydactyly and brachydactyly of both hands and feet are common. Less likely, partial syndactyly, fifth finger clinodactyly and sandle gap can also associate. In our patient there is one with polydactyly and the other without; the one with polydactyly also has sandle gap⁹. In 44% of cases with BBS has mental retardation i.e, an IQ <79. Our patient has an IQ of 54 AND 62 respectively⁹. In about 46% renal structural anomalies can be seen. Renal dysfunction has also been reported. In this case, there is hypogonadism, obesity, mental retardation, retinal degeneration in both the cases along with polydactyly in one of the child. Associated night blindness, facial dysmorphism also adds to the diagnosis of BBS. USS abdomen has been to rule out genital and renal abnormalities, which was normal for both this child. Renal function parameters were normal⁹.

Conclusion

As there are four of five major characteristics in both the children, we could conclude that it is Bardet Biedl syndrome. In addition to these major characteristics there are many minor characteristics also like global developmental delay, mild dysmorphism etc.

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