Bardet Biedl Syndrome

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Abstract

This is a case report of Bardet-Biedl syndrome, diagnosed in a 8 year old girl presented with obesity. She had polydactyly, mild mental retardation, retinitis pigmentosa in both eyes, with hypoplastic uterus, fallopian tube and ovaries. She was born to a consanguinous marriage and had a family member with same features.

Key words: Bardet biedl syndrome, Polydactyly

Introduction

Bardet-Biedl syndrome is a rare autosomal recessive disorder with cardinal symptoms of central obesity, retinal dystrophy, polydactyly, mental retardation and hypogonadism and renal dysfunction.[1] The frequency of the syndrome is estimated to be 1:1,60,000[2]. Less than 15 cases have been reported from India[3]. The incidence is higher in populations with a high level of consanguinity. Clinical diagnosis is based on the presence of 4 of the 5 cardinal features[1].

The authors present a classical case of Bardet Biedl syndrome presenting to paediatrics outpatient department with childhood obesity, mild mental retardation with polydactyly of all four limbs, and later on found to have retinitis pigmentosa and hypoplastic uterus, fallopian tubes with bilateral small ovaries with mild structural anomaly of both kidneys. There was history of consanguinous marriage in parents and presence of similar features in a relative.

The Case

Our patient was a eight year old girl, born to consanguinuos marriage, with birth weight 3 kg and length 50 cm, presented to paediatric outpatient department with obesity and night blindness. she had a second degree relative with similar features in her family. Physical examination revealed presence of central obesity with polydactyly and brachydactyly of both hands and feet. ophthalmic examination revealed atypical retinitis pigmentosa with low visual acuity (6/60 both eye). ultrasonographic examination showed hypoplastic fallopian tube and uterus with bilateral small ovaries. There was mild renal anomaly. She was 125 cm tall and weighted 50 kg with resulting BMI 32. Her IQ was 70. MRI brain was normal. History and physical examination suggested that the patient was suffering from a rare cause of childhood obesity, Bardet Biedl syndrome. Parents of the patient was counselled and advised for regular follow up for development of renal disease and progression of visual problem.

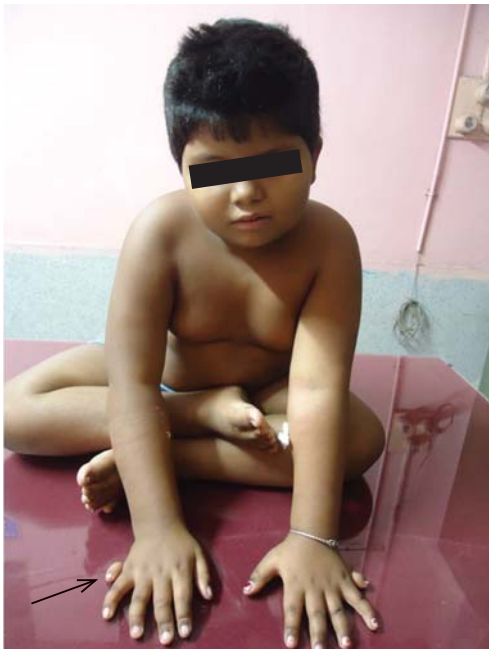

Fig 1: Polydactyly, brachydactlyly of hands

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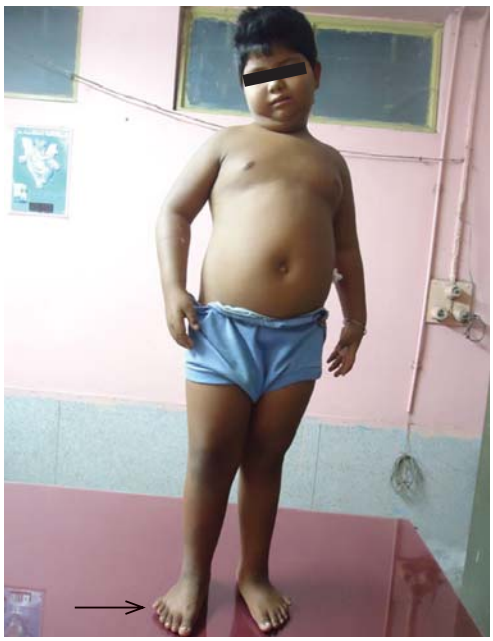

Fig 2: Central obesity and polydactyly of feet

Discussion

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive disorder with clinical and genetic heterogeneity. This syndrome was first described by Laurence and Moon in 1866 and additional cases were described by Bardet and Biedl between 1920 and 1922[4]. BBS was formerly grouped with Laurence-Moon-Biedl syndrome, but today is considered as a separate entity. It has autosomal recessive inheritance characterized by retinal dystrophy or pigmentary retinopathy, polydactyly, obesity, mental retardation, hypogonadism and hypogenitalism in males, amenorrhea in females and structural abnormalities or functional impairment of kidney[5,6,7]. Other clinical features include speech disorder, brachydactyly, developmental delay, polyuria and polydipsia, ataxia, poor coordination/clumsiness, diabetes mellitus, left ventricular hypertrophy, hepatic fibrosis, and renal hypoplasia/dysplasia. Bardet-Biedl syndrome is both phenotypically and genetically heterogeneous.

Bardet-Biedl syndrome is genetically heterogeneous, with 12 BBS genes (BBS1â€“12) identified to date[3-10]. Although the cellular mechanisms that underlie BBS remain unclear, it is now evident that all of the known BBS proteins are components of the centrosome and/or basal body and have an impact on ciliary transport[8]. The BBS1, BBS2, BBS3 and BBS4 genes contribute to ocular phenotype[9,10]. The BBS10 gene encodes a vertebrate-specific chaperonin-like protein[11] The BBS5-9 and BBS11 genes are expressed in adipose tissue [12,13]. The BBS12 gene is vertebrate specific and, together with BBS6 and BBS10 genes, defines a novel branch of type-II chaperonin superfamily[14].

Our patient presented with features of central obesity, mild mental retardation, polydactyly and brachydactyly, atypical retinitis pigmentosa, hypoplastic uterus, fallopian tubes and bilateral small ovaries with mild renal structural anomaly. Her parents was counselled and advised for regular follow for renal problem, visual difficulty and pubertal problems.

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