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Case Report

Bardet-Biedl Syndrome-Revisited

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Abstract

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive genetic disorder characterised by rod-cone dystrophy, polydactyly, central obesity, mental retardation, hypogonadism, renal dysfunction, developmental delay, speech deficit, brachydactylic or syndactyly, dental defects, congenital heart diseases, etc. Our patient had six primary and six secondary features of the disease (there could be more features but the patient did not undergo the haematological investigations ordered).

Keywords: *Genetic; Ocular; Systemic*

Introduction

Bardet-Biedl syndrome (BBS) is a rare autosomal recessive genetic disorder characterised by rod-cone dystrophy, polydactyly, central obesity, mental retardation, hypogonadism, renal dysfunction, developmental delay, speech deficit, brachydactylic or syndactyly, dental defects, congenital heart diseases, etc.

Case Report

A 35-year-old male (Figure 1) patient was brought by his brother to the department of ophthalmology with a history of night blindness for the past few years. He was an old diagnosed case of developmental delay and mental retardation. There was no other significant medical, surgical, family, traumatic or other drug intake history. Ocular examination was carried out and his visual acuity was perception of light in both the eyes (response was elucidated with great difficulty), pupillary reactions were sluggish in both the eyes, visual axis revealed exotropia though the type could not be assessed because of poor fixation and intraocular pressure was normal bilaterally. Fundus examination of both the eyes showed bilateral retinitis pigmentosa (Figure 2a and 2b).

Positive findings on his systemic examination was the presence of hexadactyly of right feet (Figure 3), weight of 100 kg with a body mass index of 29 Kg/m^2 (obesity), and hypogonadism. Ear examination revealed bilateral chronic suppurative otitis media with conductive deafness. Other positive findings were speech deficit and ataxia. Rest of his systemic examination was within normal limits. Ultrasound examination abdomen revealed fatty liver. The patient refused to undergo any haematological and cardiovascular investigations. The patient was attached to various specialities for any likely treatment from their side. We are still awaiting his follow up.

Figure 1

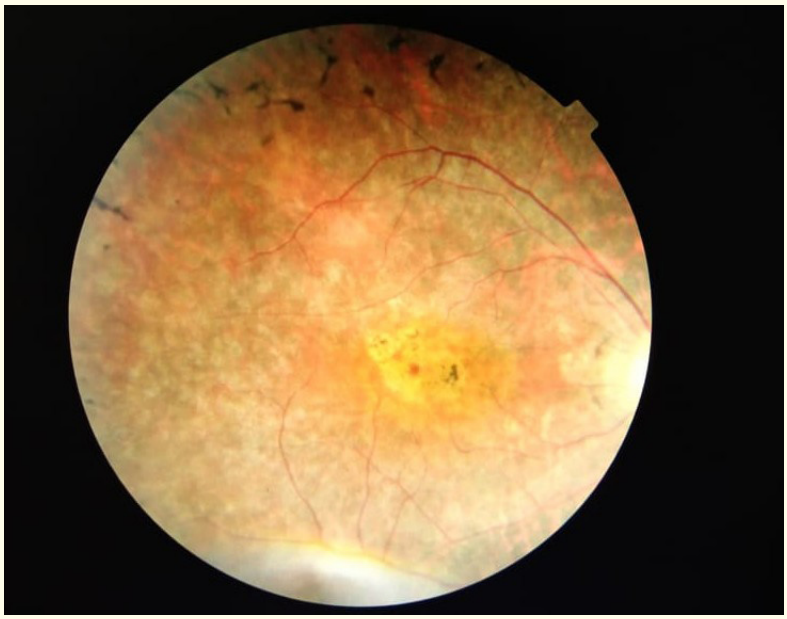

Figure 2a: *Right Eye*

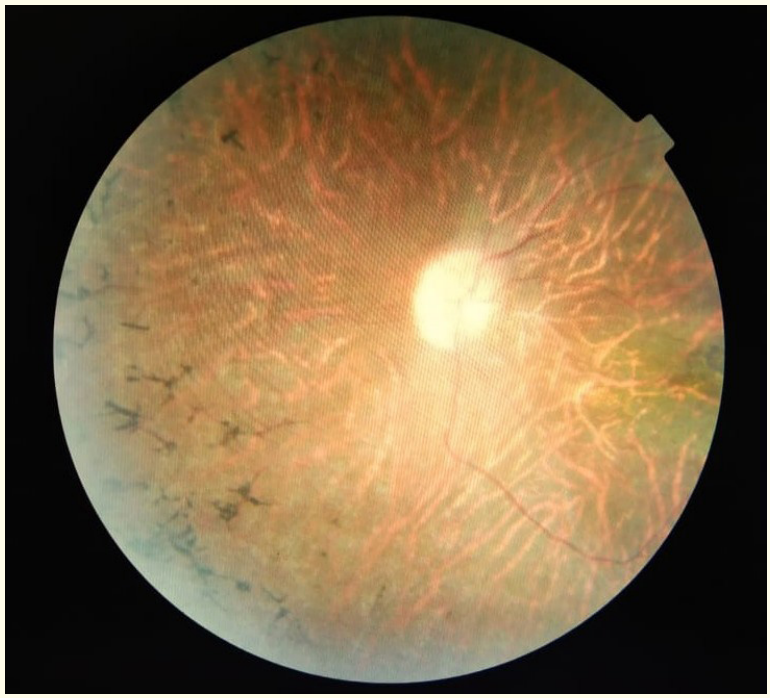

Figure 2b: *Left Eye*

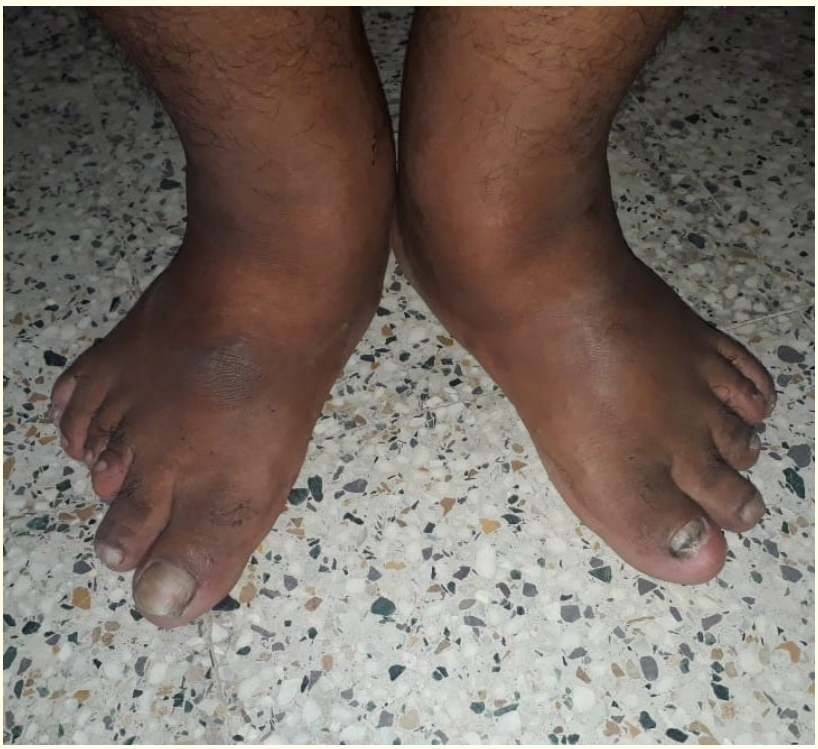

Figure 3

Discussion and Conclusion

The frequency of the BBS syndrome is estimated to be 1 in 1,60,000. Only few cases of BBS have been reported from India. BBS is named after Georges Bardet and Arthur Biedl. The first known case was reported by Laurence and Moon in 1866. Laurence-Moon-Bardet-Biedl syndrome is no longer considered as a valid term as a patient of Laurence-Moon syndrome had paraplegia but no polydactyly and obesity, which are the key elements of the BBS [1]. There are 6 primary features of BBS, i.e. rod-cone dystrophy, polydactyly, obesity, genital abnormalities, renal defects, and learning difficulties. Secondary features include developmental delay, speech deficit, brachydactyly or syndactyly, dental defects, ataxia or poor coordination, olfactory deficit, diabetes mellitus, congenital heart disease, hypertension, liver abnormalities, bronchial asthma, otitis, rhinitis, craniofacial dysmorphism, etc. For diagnosing BBS, a patient must have at least 4 out of 6 primary features of the disease. If only 3 primary features are detected, 2 secondary features are required to confirm the presence of BBS [2].

The frequency of the syndrome is estimated to be 1:160,000. Less than 15 cases have been reported from India. The incidence varies among world populations with high incidence of 1 in 13,000 in the isolated populations of Newfoundland and Kuwait. Chronic renal dysfunction is a major cause of morbidity and mortality. One of the stressful periods among these patients is during puberty and hence it is beneficial to these patients to receive guidance from an expert [3]. Other ophthalmological findings include nystagmus, strabismus, high myopia, cataract, and glaucoma [4]. BBS is a rare, genetic multi-system disorder; a ciliopathy secondary to the basal body dysfunction. Mutations in 14 genes are known to be associated with BBS: BBS1, BBS2, ARL6/BBS3, BBS4, BBS5, MKKS/BBS6, BBS7, TTC8/BBS8, B1/BBS9, BBS10, TRIM32/BBS11, BBS12, MKS1/BBS13 and CEP290/ BBS14 [5]. Investigations recommended are- ERG (Electroretinogram)/

VEP (visual evoked potentials), Renal ultrasound, IVP (intravenous pyelography), Echocardiography, Speech assessment and therapy. Semi-annually: Urine analysis. Annually: Blood pressure, Serum urea and creatinine levels, Blood sugar, Lipid profile, Liver function tests [6].

Differential diagnosis includes McKusick-Kaufman syndrome, Alström syndrome, Joubert syndrome, Senior-Løken syndrome, Leber congenital amaurosis, Biemond syndrome type II. The systemic management which should be carried out is as follows. Ophthalmologic assessment to determine visual acuity, field deficits, refractive errors and fundus examination and use of low-vision aids, examination of the genitalia, calculation of body mass index (BMI), ruling out nephrogenic diabetes insipidus, cardiac evaluation, developmental assessment, blood sugar, lipid profile, thyroid and liver functions, tests of pituitary function may be done for assessing fertility and development of secondary sex characteristics, hearing evaluation, dental evaluation to assess for hygiene, dental crowding, and hypodontia, neurologic examination for ataxia plus cerebellar functions, and genetic analysis,. To manage obesity, diet, exercise, and behavioural therapy is advocated. Hypercholesterolemia and diabetes mellitus should be treated as in the general. Speech therapy should be offered in those with speech delay. Renal transplantation has been successful, although the immunosuppressants used following transplantation can compound the weight problem. Surgical correction of hydrocolpos, vaginal atresia, or hypospadias may be warranted. As children approach puberty, gonadotropin and sex hormone levels should be monitored to determine if hormone replacement therapy is indicated. The earliest and most common intervention for polydactyly is removal of the accessory digit. Treatment of cardiac abnormalities is the same as for the general population. Dental extractions are appropriate as required for dental crowding. Prompt treatment for acute and chronic otitis media should be considered [7].

Potential future therapies, which are likely to benefit patients with BBS in the future are Genetic Therapies, Readthrough Therapy, Exon Skipping Therapy, Genome Editing, Targeted Therapies (intravenous administration of a melanocortin receptor agonist), Drug Repurposing (rapamycin may be a candidate for rectifying the renal cystic phenotype) [8].

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