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A rare case of Bardet-Biedl Syndrome

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Abstract:
Bardet – biedl syndrome(BBS)is a raregeneticdisorderwith autosomal recesiiveinheritance. Its incidence ranges from 1 :1,60,000in Northern Europe to 1 :13,500 in African population. It was first describedin1866 by ophthalmologist Lawrence and Moon and latersimilarcaseswerereported by Bardet and Biedl. The two syndromes Lawrence- Moon and Bardet – Biedl are considered to have overlappingphenotypes and werepreviouslytogethercoined as Lawrence-Moon-Bardet- Biedl syndrome. BBS typicallydevelopes over the first decade of life and presents in latechildhood or earlyadulthood. The variousclinicalfeaturesinclude post axial polydactylyassociatedwithsyndactyly and /or brachydactyly, obesity, rod-cone dystrophies, renalabnormalities, hypogonadism, developmentaldelay and cognitive impairement and speech delay. A case of BBS usually presents to the ophthalmologist with complaints of diminution of night vision. They are seen to present as atypical retinitis pigmentosa where rod involvement occurs first followed by cone involvement which leads to loss of colour vision and central vision as well in the later stages of the disease. The diagnosis can be made by the typical clinical features and electroretinography is the investigation of choice to detect changes in retina before symptomatic onset of disease. Here we present a case of a 10 year old boy who presented to us with typical features of BBS that is rod conedystrophy, polydactyly, obesity, hypogonadism and developmental and speech delay.

Key Word*: Bardet Biedl syndrome, autosomal recessive, rod cone dystrophy*

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I. Introduction

Bardet – Biedl syndrome (BBS) is a rare genetically heterogeneous disorder with autosomal recessive inheritance. [1] It is classified under a group of disorders called cilliopathies, where there is a defect in the primary cilia responsible for sensory perception and various signaling pathways.[1] This syndrome was first described in 1866 by two ophthalmologists Lawrence and Moon.[2] Later a very similar condition was reported by Bardet [3]and Biedl [4]. This condition is now considered to have two entities with overlapping phenotype- the Lawrence Moon syndrome and the Bardet Biedl syndrome. The Bardet- Biedl syndrome has a spectrum of clinical presentation. Most patients present in late childhood and early adulthood.[5] The only dysmorphic feature at birth is post axial polydactyly, which may involve all four limbs, or the upper or lower limb and this may be associated with brachydactyly and/or syndactyly.[5] Patients mostly present to the ophthalmologists in their early or mid-teens with diminution of night vision. BBS leads to development of Rod-cone dystrophy [6] and presents as an atypical retinitis pigmentosa with early macular involvement [7]. Diminution of night vision is followed by development of photophobia and loss of central vision and colour vision.[6] Other ophthalmological manifestations may include cataract and refractive errors.[5].Other major clinical findings include obesity, hypogonadism, developmental delay and cognitive deficit.[8] Renal abnormalities can be major cause of morbidity and mortality[9]. 60 % of cases have speech deficit.[10]We present one such rare case of Bardet- Biedl syndrome that presented to us with defective night vision.

II. Case report

A 10 year old male child presented to our paediatric ophthalmology outpatient department with complaints of defective vision since 3 monthswhich was more in the evening and night. The patient had history of use of glasses since last 2 years. There was no history of trauma or eye surgeries. The patient was a second child born out of a consanguineous marriage. He was born full term by normal delivery and perinatal history was insignificant. The parentscomplaint that the child had motor developmental delay and speech delay for which he was on speech therapy. The child did not do very well in school. On general examination of the child it was seen that the patient had postaxial polydactyly in three limbs (figure 1). The child had a round face and

appeared obese for his age with obesity being truncal (figure 2). On measuring, the childs height was 144 cm and his weight was 70 kg, BMI being 33.76. The child had underdeveloped penis and testes characteristic of hypogonadism. Rest of the systemic examination appeared to be normal. On ophthalmological examination, best corrected visual acuity was 6/12 in both eyes and the anterior segment appeared to be within normal limits in both eyes. Fundus examination was done after instillation of dilating drops. The optic disc appeared pale and there was attenuation of blood vessels in both eyes giving a picture of atypical retinitis pigmentosa. Bulls eye maculopathywas seen in both eyes (figure 3). A full field electroretinography was done and the result was consistent with rod cone dystrophy. With this, a provisional diagnosis of Bardet – Biedl syndrome was made and the patient was evaluated to rule out other systemic presentations of the same. Fasting and post prandial blood sugar was within normal limit. Renal function test and liver function test revealed no anomaly. Electrocardiography and echocardiography suggested a normally functioning heart. The patient was given the best correcting glasses as at the present, glasses showed significant improvement. The parents were explained about the grave visual prognosis of the disease and were advised for periodic follow up. Availability and need for low vision aids later in the course of disease was also explained to the patient. Patient was advised for weight loss. To add to this, the parents were advised for repeated visits to the paediatrician to monitor blood sugar levels and renal function owing to the risk of Diabetes mellitus and renal dysfunction that may develop later. The patient's siblings were evaluated and were found to be normal. Genetic counseling was done and parents were asked for prenatal testing in case of any future pregnancies.

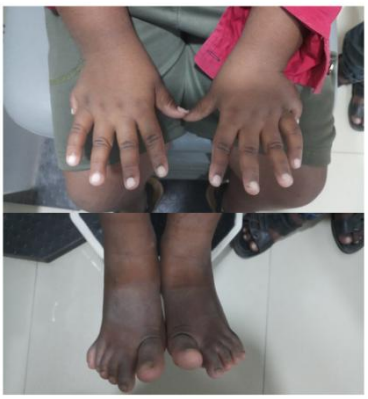

Figure 1- polydactyly in all limbs

Figure 2- central obesity

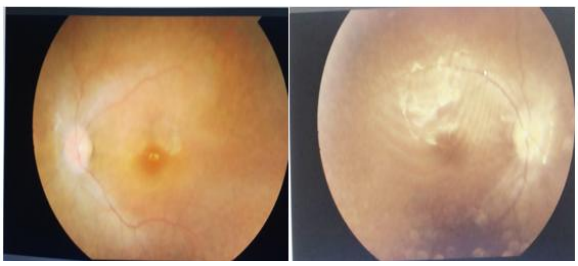

Figure 3- Pale disc, attenuated vessels and bull's eye maculopathy in both eye fundus

III. Discussion

BBS is a heterogeneous autosomal recessive disorder which is seen to be more prevalent in consanguineous births.[1]

More than 20 different genes have been identified in this condition (BBS 1 – BBS2) have been identified in this condition, out of which BBS 1 and BBS 10 are responsible for most cases.[11]The patients present with wide range of clinical features, out of which rod –cone dystrophy is the most common and is seen in 93 % of cases.[1]As in our case patients often present in pre or early teens to the ophthalmology OPD with complaints of defective vision. BBS is known to have a poor visual prognosis, and patients become legally blind by the 2nd or 3rd decade of life.[12]Typical clinical phenotype like obesity, polydactyly and hypogonadism along with electroretinography form important diagnostic modality to clinch a diagnosis of Bardet –Biedl syndrome in a case of rod cone dystrophy. Apart from this patients with BBS have motor developmental delays, cognitive impairment,speech delay. Patients are prone to develop Diabetes mellitus type 2 and renal dysfunction. The management of a case of BBS include best glasses, low vision aids when needed and mobility training[5]. Weight reduction is important. Speech therapy andvocational training are other important management modalities. Apart from this regular monitoring of blood sugar and renal function assessment is important.

IV. Conclusion

BBS develops over the first decade but even in presence of other phenotypical abnormalities like polydactyly, obesity and hypogonadism, onset of defective night vision may be the first clinical presentation of BBS which compels the patient to visit the doctor. As, BBS encompasses a wide range of systemic manifestation and there is a risk of diabetes mellitus, renal abnormalities and cardiac abnormalities, it becomes necessary that such patient is thoroughly examined and investigated and treated likewise. To date there is no effective treatment for rod cone dystrophy associated with this condition. Low vision aids are the only used modalities which fail largely at the later stages of the disease. A few years back, Simons *et al*. described a gene

therapy by adeno-associated virus-mediated *Bbs4* delivery in mouse models.[13]This may be the first step towards developing a treatment for BBS related retinal dystrophy. As on date, other associated systemic anomalies are treated symptomatically along with periodic follow ups.

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