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Pigmentary Retinopathy in Bardet Biedl Syndrome Patient, A Case Report

Case Report

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Abstract

In this case report, we report our approach in a rare case of Bardet Biedl syndrome in 8 years old male patient, found to have poor vision, nystagmus, exotropia, pigmentary retinopathy, polydactyly, truncal obesity, hypogonadism and hypothyroidism. Periodic examination and follow-up are important to prevent disease associated morbidity and mortality due to serious systemic associations. We stress on genetic evaluation especially in consanguineous populations to prevent the recurrence of the disease in future generations.

Keywords: Bardet Biedl; Truncal Obesity; Polydactyly; Retinitis Pigmentosa.

Introduction

Bardet-Biedl syndrome (BBS) is a rare ciliopathic autosomal recessive genetic disorder that affects many organ systems. The incidence of the syndrome is estimated to be 1:160,000. The prevalence is much higher in some populations with a high rate of consanguinity [1, 2]. A previous study published from in Saudi Arabia on 19 patients representing seven Saudi families, found a remarkably high frequency of consanguinity in their patients (100%) compared to the population average (56%)[3]. The main clinical features of Bardet-Biedl syndrome have been reported as retinal dystrophy (93%), obesity (91%), cognitive deficit (87%), hypogonadism (74%), and polydactyly (73%)[4]. Children with Bardet-Biedl syndrome have a poor visual prognosis with progressive loss of visual acuity that appears early in the first decade of life [5]. In this case report, we present and describe our approach in a case of pigmentary retinopathy due to Bardet-Biedl syndrome.

Case Presentation

An 8-year-old male patient presented to the clinic with his father complaining of poor vision since second year of life with nyctalopia and nystagmus. No other ophthalmological or systemic symptoms were noted from the parents. He is a product of full term, healthy pregnancy, spontaneous vaginal delivery with no complications or neonatal intensive care unit admission. Medically, he is a known case of hypothyroidism well controlled bythyroxine. Other review of systems were negative. The family history revealed epilepsy in one uncle and no cardiac, renal, hematological diseases. There is positive consanguinity between the parents, but no relatives were known to have a similar condition or ophthalmological diseases. Other siblings are normal.

We referred the case to a pediatrician to do full systemic evaluation and workup. Their evaluation was as follows: The patient was alert, conscious, active. His vital signs were blood pressure 110/64 mmHg and pulse 110 beats/mins. He is short in stature with truncal obesity and his weight was 62.3 kg and height 102cm. Facial features showed deep-set eyes and low-set ears. He has short arms, brachydactyly in hands and polydactyly in his feet. Respiratory system, cardiovascular, abdominal, and neurological examination were normal. Examination of genitalia revealed hypogonadism.

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Ophthalmological examination showed corrected visual acuity (VA) of6/60 in the right eye (OD) and counting fingers at 3 meters in the left eye (OS). Refraction is (OD) -0.50-2.50X10,(OS) -2.25-2.50X170, orthoptic examination showedExotropia (XT) of 20prism diopter (PD) for near and distance, Color vision was completely absent. Extra-ocular muscles movements evaluation showedfull movements of both eyes with nystagmus which was jerky, very high frequency, short amplitude, horizontal, uniplanar and increase in extreme gazes. Pupils were round, regular, reactive. He has a good red reflex. Lids and lashes were within normal limits, conjunctiva was quiet, cornea was clear, iris within normal limits, anterior chamber was deep and quiet. Dilated fundus examination showed bilateral temporal optic nerve pallor, with cup-to-disc ratio of 0.4,retinal thinning in the macula andpigmentary retinopathy with attenuated blood vessel. (Figure1) (Figure2).

We wanted to refer the patient for genetic evaluation and his family for screening, but it was not available in our institute and patient's parents were not interested to do that in other institute due to financial limitations.

Discussion

Bardet-Biedl syndrome (BBS) is a genetic heterogeneous disorder characterized by a broad spectrum of clinical features described by Bardet Biedl in 1920 [6, 7]. BBS can result from mutations in ≥20 different genes [1]. Some recent genetic studies that defined the syndrome by specific genetic mutations. Eleven genes are discovered to be associated with this syndrome, and those are BBS1, BBS2, ARL6/BBS3, BBS4, BBS5, MKKS/BBS6, BBS7, TTC8/BBS8, B1/BBS9, BBS10, TR1M32/BBS11 [8].

Diagnosis is established by clinical findings. Beales et al. have described the primary and secondary features which are illustrated in Table (1). The Primary clinical features include retinitis pig-

Figure 1. Fundus photo showing: bilateral temporal optic nerve pallor, with cup-to-disc ratio of 0.4, retinal thinning in the macula and pigmentary retinopathy with attenuated blood vessel.

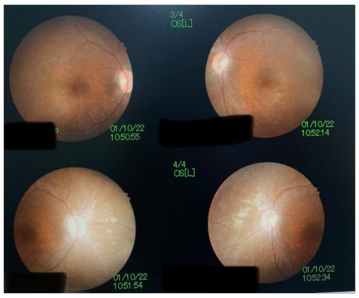


Figure 2. Optical coherence tomography (OCT) of macular area in both eyes showing retinal thinning.

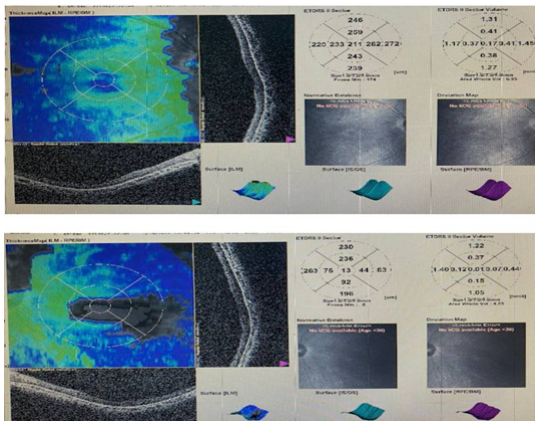
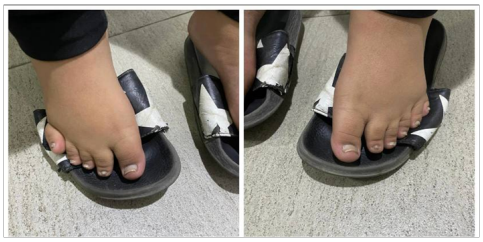


Table 1. Bardet Biedl Syndrome (BBS) diagnostic criteria established by Beales et al.

Primary Features	Secondary Features
1. Rod-cone dystrophy	1. Speech disorder/Delay
2. Obesity	2. Strabismus/cataracts/astigmatism
3. Polydactyly	3. Brachydactyly/ syndactyly
4. Learning Disabilities	4. Developmental delay
5. Hypogonadism in males	5. Nephrogenic diabetes insipidus
6. Renal Anomalies	6. Ataxia/poor coordination/imbalance
	7. Mild spasticity
	8. Dental crowding/hypodontia/small roots
	9. Left ventricular hypertrophy/congenital heart disease
	10. Hepatic fibrosis

Figure 3. External photo of both feet showing post-axial polydactyly.



mentosa (RP), polydactyly, hypogonadism, obesity, intellectual disabilities, and renal abnormalities. Secondary clinical features include speech disorders, strabismus, brachydactyly or syndactyly, developmental delay, polydipsia-polyuria (diabetes insipidus), ataxia, spasticity, diabetes mellitus, dental crowding or hypodontia, congenital heart diseases, and hepatic fibrosis [1]. The presence of four primary features or three primary features plus two secondary features is diagnostic [6]. Our patient had four primary features: Retinitis pigmentosa, Obesity, Polydactyly, and Hypogonadism, thus fulfilling the diagnostic criteria.

Among all these features, Retinal dystrophy is the most common and penetrant feature of BBS, making this disorder the second most common syndromic retinal degeneration behind Usher syndrome [9, 10]. Retinitis pigmentosa (RP) is the name given to a group of genetic eye diseases that affect the retina and cause gradual, permanent loss of vision due to progressive photoreceptor dysfunction, followed by photoreceptor cell death [5]. RP is the most common inherited retinal degeneration, with an estimated worldwide prevalence of 1:4000 [7].

BBS patients typically develop retinitis pigmentosa (RP) symptoms in the first decade of life and often reach legal blindness between the second and third decades of life. Night blindness (nyctalopia) is the most common initial visual symptom and is usually first noted around 8.5 years [10]. In our patient, the family start to notice a poor vision at night with nystagmus since birth.
The second prominent feature of BBS is Obesity which usually begins in childhood. Majority of cases exhibit symptoms within the first year of life. In our reported case, he developed truncal obesity in childhood. Hypogonadism is reported more frequently in BBS males than females [3, 6]. Which is manifested in our patient.

Postaxial polydactyly has been frequently reported in different ciliopathies, especially in BBS. Our patient has polydactyly in both hands [11]. Figure (3).

Conclusion

The management of Bardet Biedl Syndrome (BSS) is challenging and requires multidisciplinary efforts from pediatricians, orthopedic surgeons, pathologists, audiologists, ophthalmologists, nephrologists, genetics and other healthcare professionals. Periodic systemic and ophthalmologic evaluations is the key to prevent the disease associated morbidity and mortality. Early intervention is essential in ensuring that children with Bardet Biedl syndrome reach their highest potential. Genetic evaluation and screening is helpful especially in consanguineous parents to prevent the recurrence of similar disease in future off-springs.

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