Incomplete presentation of Bardet–Biedl syndrome with atypical retinitis pigmentosa

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Abstract

Typical retinitis pigmentosa (RP) is less commonly seen here than atypical presentations, such as cone-rod dystrophy, RP sine pigmento, and retinitis punctata albescens. We report a patient presenting with incomplete systemic manifestations of Bardet–Biedl syndrome and atypical RP, with possible differential diagnoses of overlapping syndromes. It is prudent for an ophthalmologist to suspect syndromic RP in patients presenting with pigmentary retinopathy and systemic abnormalities, as they require a multisystem approach to reduce morbidity.

Keywords: Bardet–Biedl syndrome, Retinitis pigmentosa, Syndromes

INTRODUCTION

Bardet–Biedl syndrome (BBS) is a rare, autosomal recessive ciliopathy characterized by retinal pigmentary dystrophy, postaxial polydactyly, renal dysfunction, learning difficulties, hypogonadism, and obesity.[1] We report a patient presenting with atypical retinitis pigmentosa (RP) and systemic manifestations suggestive of BBS.

CASE REPORT

A 17-year-old female presented to our clinic with complaints of diminution of vision and outward deviation of the left eye since birth. Birth history, birth weight, and developmental milestones were normal. General examination showed normal BMI, short stature, postaxial polydactyly, and normal intelligence. Her unaided vision was 6/6p in the right eye and 6/12p in the left eye. Hirschberg test showed a left exotropia. Inferior oblique overaction and nystagmus of both eyes were seen. Anterior segment evaluation of both eyes was within normal limits. Fundus examination of both eyes showed pale discs with a cup disc ratio of 0.2, RPE alterations at the posterior pole, arteriolar attenuation, and few bony spicules in the mid periphery. Color vision testing on Ishihara charts was 0/17 in both eyes, and Humphrey visual fields showed severely depressed fields in both eyes. She was diagnosed with BBS with atypical RP.

DISCUSSION

BBS is a multisystem ciliopathy of which retinal disease is the most penetrant feature.[2] It is the second most common syndromic retinal degeneration after Usher syndrome.[3] The retinal disease shows early macular involvement with a spectrum from subtle maculopathy to rod-cone dystrophy with peripheral bony spicules to global retinal degeneration.[4-6] Our patient had pigmentary changes at the macula with sparse mid-peripheral bony spicules [Figure 1a-e].

Loss of rods presents with night blindness and decreased peripheral vision. Cones are lost with progressive rod dysfunction, leading to reduced visual acuity and loss of color discrimination. Our patient had postaxial polydactyly but did not have other features of BBS such as obesity, hypogonadism, renal dysfunction, or cognitive impairment. Cognitive impairment is the most variable phenotype of BBS, with some patients reported to have

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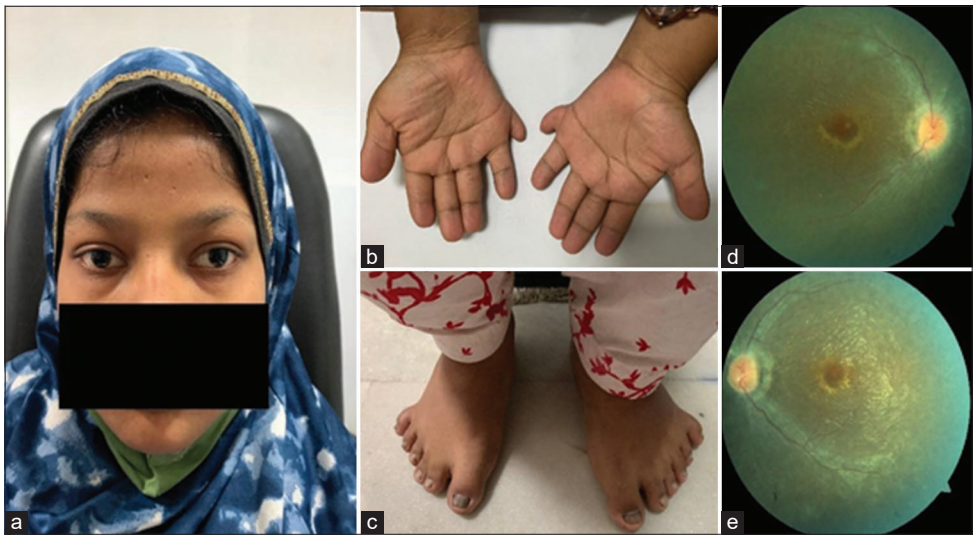

Figure 1: (a) Outward deviation of the left eye (b and c) Postaxial polydactyly (d and e) Fundus examination of both eyes (image d being of the right eye, image e being of the left eye) showed pale discs with cup disc ratio of 0.2, RPE alterations at the posterior pole, arteriolar attenuation, and few bony spicules in the mid periphery

Table 1: Summary of the possible differential diagnoses of Bardet–Biedl syndrome in contrast to this case[8]

S. No	Disorder	Clinical presentation in our patient	
		Overlapping with disorder	Different from disorder
1.	Bardet–Biedl syndrome	Rod-cone dystrophy Postaxial polydactyly	No obesity, hypogonadism, or renal dysfunction Preserved cognitive function
2.	Joubert syndrome	Retinal degeneration polydactyly Absence of central obesity and hypogonadism	No hypotonia, breathing abnormalities, kidney or liver disease
3.	Alström syndrome	Rod-cone dystrophy Normal cognitive functions	No obesity, hypogonadism, or renal dysfunction Presence of polydactyly
4.	McKusick–Kaufman syndrome	Postaxial polydactyly Absence of obesity and developmental abnormalities	No heart or genitourinary disease Presence of retinal disease
5.	Leber's Congenital Amaurosis	Retinal degeneration	Presence of multi-system manifestations

higher intellectual abilities.[4,6] A close differential due to the presence of retinal degeneration and polydactyly, and the absence of obesity and hypogonadism in the presentation, was Joubert syndrome.[7] Our patient did not have hypotonia, ocular motor apraxia, or breathing abnormalities seen with Joubert syndrome. We have summarized the possible differentials and their manifestations in [Table 1].

Renal dysfunction is a major cause of morbidity and mortality and should be monitored in patients with BBS.[9] We report this case as an incomplete presentation of BBS without ruling out a possible overlap between the syndromes.

CONCLUSION

Although rare, Bardet–Biedl syndrome should be suspected when a patient presents with pigmentary retinopathy and multisystem manifestations. Due to the high penetrance of retinal disease, the ophthalmologist may be the first to encounter such patients.

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CONFLICTS OF INTEREST

None.

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None

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