Case Report

Rod-Sparing Retinopathy – Unusual Variant in Bardet–Biedl Syndrome

Sowmya Raveendra Murthy, Keerthi Gunda

Department of Pediatric Ophthalmology and Strabismus, Sankara Eye Hospital, Bangalore, Karnataka, India

Abstract

Bardet–Biedl syndrome (BBS) is a genetic disorder characterized by retinal dystrophy, obesity, postaxial polydactyly, renal dysfunction, learning difficulties, and hypogonadism. Retinal dystrophy in the form of rod-cone dystrophy is the most common diagnostic handle prompting investigation for BBS. We report a novel case of rod sparing or a cone-selective variant of retinopathy in BBS. To the best of our knowledge, this is the first report of such a case from India.

Keywords: Bardet–Biedl syndrome, obesity, polydactyly, retinitis pigmentosa, rod sparing

Introduction

Bardet–Biedl syndrome (BBS) is a genetic disorder varying in expression within and between families.[1,2] Inheritance is traditionally considered autosomal recessive, although notable exceptions exist. The syndrome is often divided into the following two entities: Laurence–Moon syndrome and BBS, but BBS is now the standard term in common usage.[3]

Newer diagnostic criteria for BBS demand the presence of four primary features or three primary and two secondary features. Primary features includes rod-cone dystrophy, polydactyly, obesity, renal anomalies, and genital anomalies with learning difficulties, whereas secondary features comprise speech delay, developmental delay, diabetes mellitus, dental anomalies, ataxia, poor coordination, congenital heart disease, anosmia, and brachydatyly. Postaxial polydactyly is common and may be the only obvious dysmorphic feature at birth.[1,3]

Retinal dystrophies in the form of rod-cone dystrophy, cone-rod dystrophy, choroidal dystrophy, or global retinal dystrophy have been described in BBS.[2] Pigmentary retinal dystrophies in varying severity of rod dystrophy including retinitis pigmentosa sine pigmento, retinitis punctata albescens, or even typical retinitis pigmentosa have been described.[3] Atypical retinitis pigmentosa with early macular involvement was also described.[4,5] Some patients with not classifiable early photoreceptor dysfunction are noted to eventually progress to a classic stage of rod degeneration.[6] However, the involvement of only cones is an unusual variant.[7]

Electroretinography is the investigation of choice and may show early changes within the first 2 years of life, although significant changes are rarely visible before the age of five.[5]

Case Report

A 12-year-old child presented with complaints of photophobia. He reported to be more at ease while reading and doing his homework at night than in daylight. He was wearing glasses since 4 years.

On examination, his best corrected visual acuity (BCVA) was 20/40, N6 (OU; both eyes) with a refraction value of (OD; right eye) −6.00 DS, (OS; left eye) −5.00 DS. Cover test showed 20–25 PD exotropia for distance and 30 PD exotropia for near.

Ocular movements showed (OU) an inferior oblique overaction of +1, with no evidence of nystagmus. Anterior segment examination was normal.

Address for correspondence: Dr. Sowmya Raveendra Murthy, Sankara Eye Hospital, Varthur Main Road, Kundlahalli Gate, Bangalore 560037, Karnataka, India.
E-mail: drsowmyamurthy@gmail.com

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Examination of the fundus showed normal discs with bull's eye lesion at the fovea and normal peripheral retina [Figure 1].

Dyschromatopsia was noted, with the child identifying only the demo plate on Ishihara charts.

A general physical examination showed obesity, polydactyly in the hands and feet (six fingers and toes), hypogonadism, and moderately low intelligence quotient (IQ).

Electroretinogram (ERG) was performed, which showed extinguished cone responses. Rod responses and combined response were normal [Figure 2].

The physical appearance of the child was similar to the features depicted in those with BBS (having four primary criteria to classify as BBS), with eye features suggestive of cone dystrophy.

There was no significant family history, with the elder sibling, parents, and cousins being normal.

Complete hormonal assay was performed, which showed cortisol, fasting blood sugar (FBS)/postprandial blood sugar (PPBS), serum calcium, and lipid levels to be within normal limits except increased plasma insulin levels. Slight increase in thyroid stimulating hormone (TSH) levels was noted. Liver function tests were normal except slightly increased gamma-glutamyl transferase levels. Body mass index was high, and the skin showed acanthosis nigricans.

Regular follow-up was done. Retinopathy remained cone selective even at the end of 2 years and 6 months.

Discussion

BBS is an autosomal recessive condition with a wide spectrum of ocular and systemic features. Our patient had primary features including polydactyly, truncal obesity, learning difficulties, and hypogonadism with cone dystrophy fulfilling the criteria to be diagnosed as BBS.

Though Alstrom disease was considered, the presence of polydactyly and normal hearing in our patient ruled out the possibility.[1]

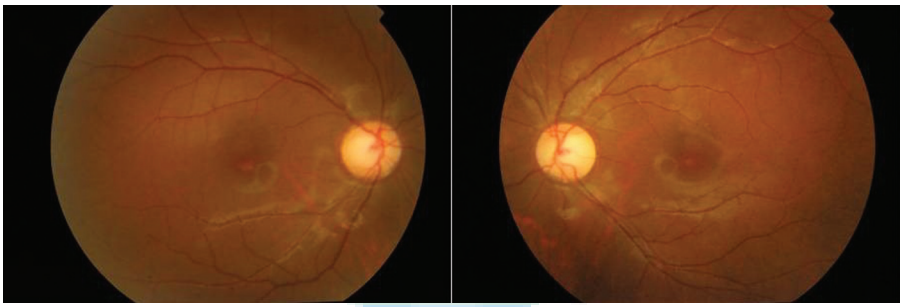

Figure 1: Fundus photographs showing bull's eye lesion at macula and normal vasculature in both eyes

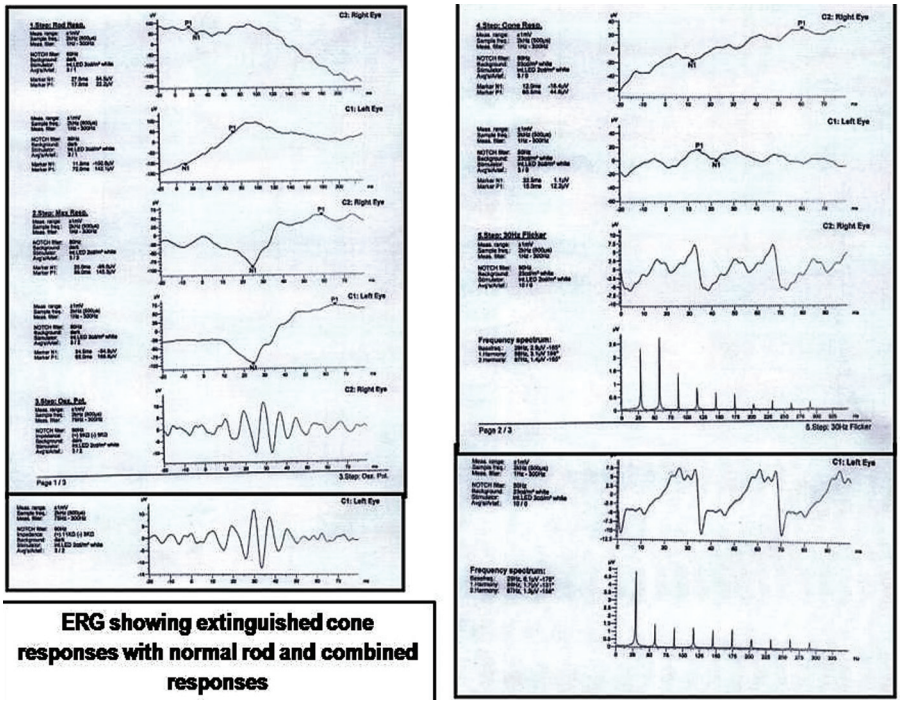

Figure 2: Electroretinogram showing extinguished cone responses with normal rod and combined responses in both eyes

The classic ocular manifestation is the gradual onset of night blindness, followed by photophobia and the loss of central and color vision. Many variants have been described, and some may develop the converse sequence of pathological events with early loss of cone photoreceptors followed by rod photoreceptors.[4]

Our patient had BBS with the involvement of cones only. The presenting features of photophobia, the loss of central vision, and defective color vision, all suggested cone involvement in our patient. Further extinguished cone response on ERG confirmed the isolated cone involvement in our patient. Similar to our case, a rod-sparing retinopathy case has been reported in a 17-year-old boy, in whom only cones were involved in initial ERG and after 4 years follow-up.[8]

Thus, we believe that our patient demonstrates a rare cone-selective variant of BBS. This is the first report of such a case from India to the best of our knowledge.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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