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A Rod-Sparing Retinopathy in Bardet-Biedl Syndrome

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Key Words
Bardet-Biedl syndrome · Rod-sparing · Polydactyly · Retinitis Pigmentosa · Electroretinography

Abstract
Bardet-Biedl syndrome is a continuum of disorders characterized by systemic and ocular findings. Retinal abnormalities typically present as diffuse photoreceptor degeneration. Here, we report a novel case that suggests a rod-sparing variant of Bardet-Biedl syndrome.

Introduction
Bardet-Biedl syndrome is a continuum of systemic manifestations that include obesity, polydactyly, hypogonadism and mental retardation [1]. The ocular examination can reveal different types and severity of pigmentary retinal dystrophy (including retinitis pigmentosa sine pigmento, retinitis punctate albescens or even typical retinitis pigmentosa) [2] as well as a combined rod-cone dystrophy characterized by global photoreceptor malfunction. However, we have identified an unusual variant of Bardet-Biedl syndrome where the retinopathy is cone selective.
Case Report

A 17-year-old Armenian boy was referred to the Neuro-Ophthalmo...
References


Fig. 1. Fundus photos of both eyes showing optic nerves with a normal appearance, and pigment mottling in the macula in the right (a) and the left eye (b). Goldmann visual fields documented bilateral central scotoma to the I4e test isopter, spanning a 10-degree radius from the center of vision both in the left (c) and the right eye (d).
Fig. 2. ERG recordings. A Ganzfeld stimulator, with bright flash intensity calibrated to 2.12 cd s/m², was used. The flash duration was 10 ms. The band pass of the amplifiers was set at 0.3–300 Hz. A 24-dB neutral density filter over a strobe lamp was presented as a dim white flash stimulus. The background of the Ganzfeld dome was then illuminated at 39 cd/m². Using the same background illumination, the bright flash was presented at 30 Hz and then sweeps were averaged. Dim scotopic (a, e) and bright scotopic (b, f) were normal, while bright photopic was unrecordable (c, g) and 30-Hz flicker showed reduced and delayed tracings (d, h), both upon first presentation as well as 4 years later (a–h).