Brief Communication

Beyond the Darkness: Navigating Oguchi Disease with a Singular Case Insight

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ABSTRACT

Oguchi disease represents a hereditary condition characterized by stationary night blindness. It has a characteristic clinical sign, the Mizuo-Nakamura phenomenon. We present a case of a 9-year-old girl who was evaluated for complaints of impaired night vision. Best-corrected visual acuity in both eyes was 20/100. Dilated fundus examination demonstrated a metallic sheen across the retina bilaterally. After a dark adaptation for 2 hours, the characteristic golden-yellow metallic sheen resolved, demonstrating the clinical phenomenon known as the Mizuo-Nakamura effect.

Keywords: Mizuo-Nakamura phenomenon; metallic sheen; Oguchi disease.

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INTRODUCTION

Oguchi disease is a hereditary condition characterized by stationary night blindness. It has a characteristic clinical sign, the Mizuo-Nakamura phenomenon, featuring the disappearance of golden-yellow retinal discoloration upon dark adaptation which returns when exposed to light. Individuals with Oguchi disease typically have difficulty seeing at night, but maintain preserved color perception, visual field, and visual acuity. Testing shows diminished or absent rod photoreceptor activity, while cone photoreceptor function remains intact.[1]

Genetic variations affecting the Arrestin or Rhodopsin kinase genes have been identified as the fundamental etiological factors underlying Oguchi disease.[2] Oguchi disease can be categorized into two distinct subtypes based on the underlying genetic etiology. Oguchi disease type 1 is linked to the presence of homozygous or heterozygous genetic variants within the SAG gene, which is situated on the long arm of chromosome 2q37.1. The S-antigen, a key component of the visual signal transduction pathway, forms a complex with Rhodopsin that has been photoactivated and phosphorylated, thus inhibiting further interactions with the activated form of Rhodopsin. In contrast, Oguchi disease type 2 is caused by mutations in the Rhodopsin kinase gene on chromosome 13q34. The GRK1 gene encodes the rhodopsin kinase enzyme, which recognizes and desensitizes photoactivated Rhodopsin, enabling the visual system to respond to new light stimuli.[3]

Oguchi disease exhibits a range of subtypes which include diffuse involvement of the entire retinal fundus, as well as more focal patterns sparing the macula, posterior fundus, peripheral retina, or far periphery.[4] Electroretinographic testing demonstrated an absence of rod-derived b-wave responses after 30 minutes of dark adaptation, in conjunction with relatively preserved a-wave amplitudes but markedly diminished b-wave amplitudes.[5]

CASE PRESENTATION

A 9-year-old young child was brought to the Pediatric Outpatient Department with the primary complaint of night blindness in both eyes. The parents reported the history of consanguineous marriage. Family medical

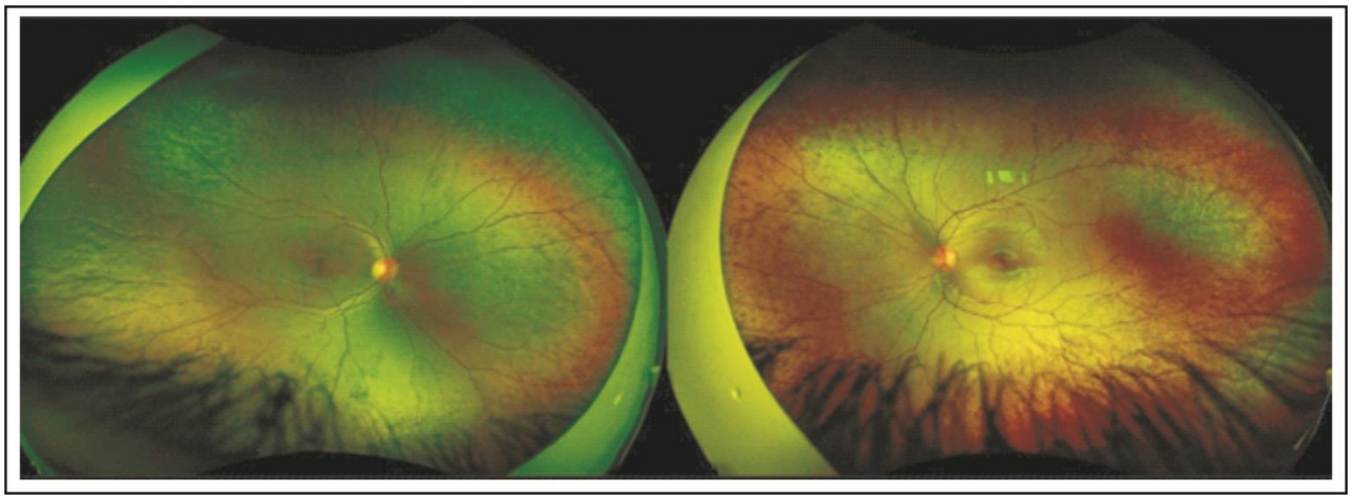

Figure 1: Colour fundus photography showing pre-dark adaptation fundus photographs of both eyes with a yellow iridescent (golden) sheen.

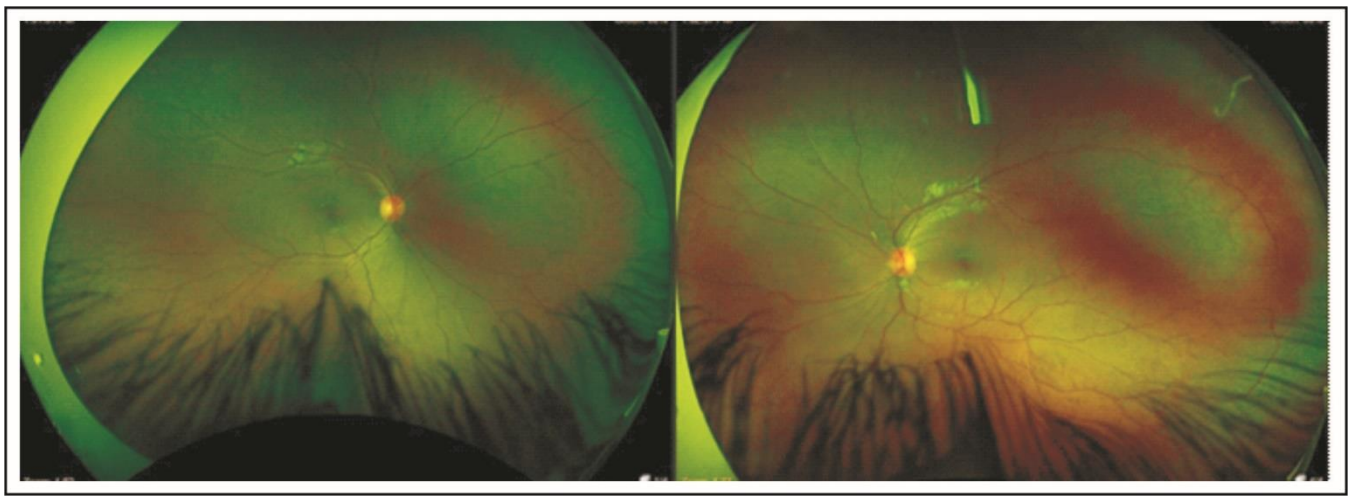

Figure 2: Colour fundus photography showing post-dark adaptation fundus photographs of both eyes highlighting the disappearance of the golden yellow metallic colour.

history was unremarkable.

Physical examination of the patient revealed no abnormalities. Ophthalmic assessment showed equal, round, and reacting pupils, as well as intact extraocular movements. The patient exhibited a best-corrected visual acuity of 20/100 bilaterally. Slit-lamp bio microscopy of the anterior segment was normal. Dilated fundus examination demonstrated a diffuse metallic sheen evident across the retinal surface bilaterally (Figure 1).

The characteristic golden-yellow metallic fundus appearance resolved following two hours of dark adaptation, demonstrating the Mizuo-Nakamura phenomenon (Figure 2).

DISCUSSION

This case report presents a patient with the rare Oguchi disease, a hereditary non-progressive night vision impairment, exhibiting the two characteristic clinical manifestations observed in this instance.[1] First, the patient presented with congenital stationary night blindness, a hallmark symptom of Oguchi disease. Second, the fundoscopic examination revealed the Mizuo–Nakamura phenomenon, where the golden-yellow metallic discoloration of the retina disappeared after dark adaptation and reappeared upon exposure to light.

The underlying pathophysiology remains uncertain, but it may involve an excess of extracellular potassium due to the impaired potassium-clearing

capacity of retinal Müller cells.[6] This phenomenon has been observed in Oguchi's disease, as well as other conditions like retinitis pigmentosa, X-linked retinoschisis and cone-rod dystrophy. Retinitis pigmentosa has progressive worsening of night vision. X-linked retinoschisis patients have foveal and peripheral splitting of retinal layers. Additionally, individuals with cone-rod dystrophy may display a bull's eye-like maculopathy.

The electroretinographic findings in Oguchi disease demonstrate characteristic reductions in amplitudes of both a-wave and b-wave. Following prolonged dark adaptation, the initially detectable rod function in Oguchi disease patients rapidly declines.[7]

The findings suggest that while dark adaptation can partially restore rod function in younger patients, the speed of this recovery is influenced by the patient's age. Deng et al, reported diminished scotopic ERG results, but more comprehensive electrophysiological assessment following dark adaptation was hindered by the patient's limited cooperation.[3] The electroretinographic assessments of our patient demonstrated findings analogous to those commonly associated with Oguchi disease.

Rishi et al, presented a case of a seven-year-old boy with a five-year history of night vision difficulties.[8] His visual acuity was 20/20 in both eyes. Fundus examination revealed a golden sheen over the posterior pole in both eyes, which disappeared after 45 minutes of dark adaptation, consistent with the Mizuo-Nakamura phenomenon. Clinical findings suggested Oguchi's disease, which was confirmed by electroretinography (ERG).

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Conflict of Interest: Authors declared no conflict of interest.

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