

Oguchi Disease or Stationary Congenital Night Blindness: A Case Report

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ABSTRACT

Oguchi disease, originally described in Japanese people, is a rare form of stationary night blindness in patients with normal visual acuity. Oguchi disease is an autosomal recessive disorder usually caused by mutation of arrestin or rhodopsin kinase. The fundus has an unusual golden-yellow colour in the light-adapted state, which becomes normal after prolonged dark adaptation called Mizuo or Mizuo-Nakamura phenomenon. The origin of the reflex is not clear. This rare disease is most commonly encountered in Japan and is presented in view of its novelty.

Keywords: Mizuo-Nakamura phenomenon, Stationary night blindness.

INTRODUCTION

Oguchi disease, originally described in Japanese people, is a rare form of stationary night blindness in patients with normal visual acuity. Oguchi disease is an autosomal recessive disorder usually caused by mutation of arrestin or rhodopsin kinase. The fundus has an unusual golden-yellow colour in the light-adapted state, which becomes normal after prolonged dark adaptation called Mizuo or Mizuo-Nakamura phenomenon.^[1,2] The origin of the reflex is not clear. This rare disease is most commonly encountered in Japan and is presented in view of its novelty.

CASE REPORT

A 16 years old healthy girl was brought by her mother for non-progressive defective vision at night in both eyes for 1 year duration. Her day time vision was normal. She did not give any history of trauma, previous ocular or systemic disease. In both eyes, uncorrected visual acuity was 6/6, N/6. Slit lamp examination of anterior segment was normal. Fundus examination of both eyes revealed grayish metallic phosphorescent sheen as shown in [Figure 1,2]. Optic disc, macula, retinal vessels were normal. When the girl was subjected to dark adaptation for 3 hours, the fundus background colour changed and became normal Mizuo-Nakamura phenomenon. Her sister has similar history of defective night vision and she also had similar findings.

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The electroretinogram (ERG) response, as illustrated in [Figure 3,4] below consists of an initial negative wave, the a-wave, followed by a second positive wave, the b-wave. The a-wave is derived from both rods and cones and b-wave is derived from inner retina, predominantly Muller cells and Optic Nerve (ON)-Bipolar cells. ERG of our patient showed a rod/max combined/Oscillatory Potentials (OPs) response, in which the b-wave amplitude was diminished in both eyes. The cone ERG was normal in both eyes.



Figure 1: OS (Right eye)

DISCUSSION

Oguchi disease first described by Chuta Oguchi in 1907, is a rare autosomal recessive disorder characterized by congenital stationary night blindness and a unique morphological and functional abnormality of the retina. Patients have non-progressive night blindness since young childhood with normal day vision, but often report improvement of light sensitivities when they remain

long in dark environment; dark-adapted study demonstrates that highly elevated rod thresholds decrease several hours later and eventually result in recovery to normal or nearly normal level³. The fundus has a diffuse or patchy appearance, silver-gray or golden-yellow metallic sheen and the retinal vessels are normal. A prolonged dark-adaptation of 3 hours or more, leads to disappearance of unusual discoloration of normal reddish appearance called Mizuo-Nakamura phenomenon.^[2,3] Oguchi's disease is also unique in the ERG responses in the light and dark-adapted conditions. B-wave amplitude is reduced in rod response and cone response is normal.^[3,4] Arrestin gene mutation in chromosome 2q in patients with Oguchi's disease may account for characteristic fundus and functional abnormality.^[3,6] As rhodopsin kinase works with arrestin in shutting off rhodopsin after it has been activated by photon of light, it has been proposed that some cases are due to defects in rhodopsin kinase.^[3]



Figure 2: OS (Left eye)

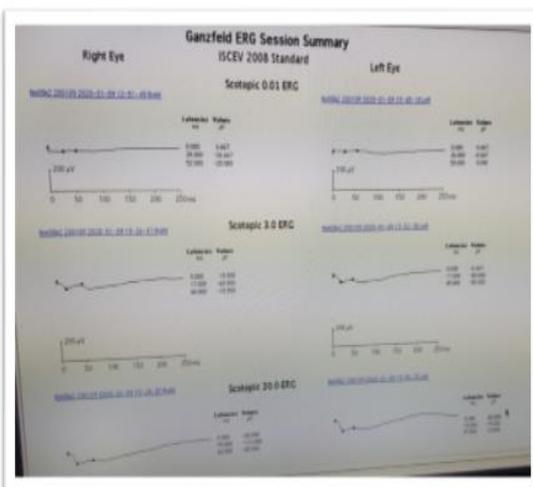


Figure 3: Electroretinogram

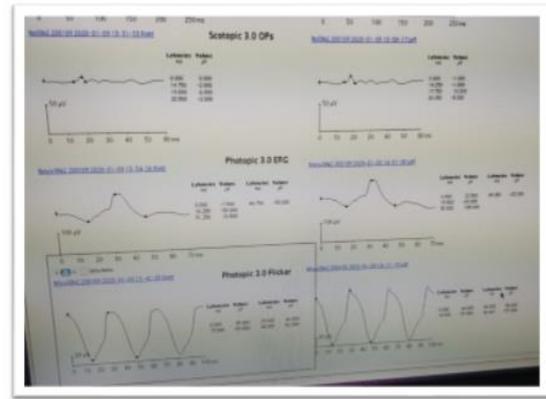


Figure 4: Electroretinogram

In our patient, the classical Mizuo phenomenon was present and the fundus discoloration returned to normal with dark adaptation. The differential diagnosis includes Stargardt's disease, female carrier of Retinitis Pigmentosa, Juvenile Retinoschisis, Progressive Cone Dystrophy, all these conditions may have fundus changes but without classical Mizuo phenomenon.

CONCLUSION

The diagnosis should be made when hemarropia is associated with typical fundus findings resolving after dark-adaptation so called Mizuo-Nakamura phenomenon. The long term prognosis in these patients is good in the absence of clinical progression.^[1,5]

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