# Electrophysiological findings in Oguchi disease

## *Alterações eletrofisiológicas na doença de Oguchi*

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### *ABSTRACT*

*To describe the electrophysiological alterations in a very rare case of Oguchi's disease. A 17-year-old italian girl complaining of night blindness underwent complete ophthalmological exams, including electrophysiological tests. Rod responses were nondetectable in full-field electroretinogram (ERG). The photopic ERG funtions, including the 30 Hz flicker ERG response was normal, while the scotopic b-wave was diminished in amplitude. The electrooculography (EOG) ratios within the normal range were 208% in the right eye and 222% in the left eye. The Mizuo-Nakamura phenomenon was present. The electrophysiological tests are important tools in Oguchi's disease diagnosis. In the present case, it's clear the non correspondance between EOG and ERG. Considering the rod function, the normal EOG ratio contrast with non-detectable rod ERG responses. More studies are necessary to understand the compless electrofuntional mecanism of the disease helping to understand the origin of the light-sensitive component of the EOG.*


***Keywords:*** *Night blindness; Oguchi's disease; Electroretinography; Electrooculography; Mizuo-Nakamura phenomenon; Case reports*


### RESUMO

Descrever as alterações eletrofuncionais em um caso raríssimo da Doença de Oguchi. Paciente do sexo feminino, italiana de 17 anos de idade se queixava de cegueira noturna. A resposta escotópica de bastonetes, do ERG era não registrável. A resposta escotópica ao estímulo branco forte demonstrava uma diminuição de amplitude da onda B. As respostas ao flicker de 30Hz e ao EOG eram dentro dos limites da normalidade. Era presente o fenômeno de Mizuo-Nakamura. Os exames eletrofuncionais são muito importantes no diagnóstico de certeza da doença de Oguchi. É nítida, no presente caso, a discordância entre EOG e ERG. Considerando a função dos bastonetes, as respostas normais do EOG contrastam com a ausência de respostas dos bastonetes em condições escotópicas no ERG. Mais estudos são necessários para entender o complexo mecanismo eletrofuncional dessa doença e melhor definir a origem dos componentes sensíveis à luz do EOG.


**Descritores:** Cegueira noturna; Doença de Oguchi; Eletrorretinografia; Eletro-oculografia; Fenômeno de Mizuo-Nakamura; Relatos de casos

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## Introduction

There are three forms of stationary night blindness (SNB): congenital stationary night blindness (CSNB), fundus albipunctatus and Oguchi disease.

Oguchi disease is a rare congenital, autosomal recessive form of SNB characterised by a peculiar greyish or yellow-greenish discoloration of the fundus[1], which reverts to normal after prolonged dark adaptation (Mizuo-Nakamura phenomenon)[2].

Patients with Oguchi disease can be classified into two types depending on the shape of the adaptation curve[1]. In type I, rod adaptation is markedly slow. Function is fully recovered after hours of dark adaptation and the curve is normal or slightly altered. In type II there is no rod adaptation, retinal changes are less clear and the Mizuo phenomenon may be absent.

Electrofunctional changes are very important in the definitive diagnosis of Oguchi disease. Electroretinography and electrooculography are useful in understanding the complex mechanism of the disease.

The aim of this paper is to describe the electrofunctional changes in a very rare case of Oguchi disease.

**Case report**

A white, female, 17-year-old Italian patient presented with complaints of night blindness. Her medical history showed no systemic diseases or previous health conditions. The patient underwent a complete ophthalmic examination including electrofunctional tests at University La Sapienza, Rome, in the year 2007. A full-field electroretinogram was performed using Metrovision equipment (MONPAK3 *Moniteur Ophtalmologique - Electrophysiologie visuelle*). Standard ISCEV stimuli were used: rod response (dim white light under scotopic conditions), maximum response (strong white light under scotopic conditions), oscillatory potential, cone response (strong white light under photopic conditions), and 30 Hz flicker. A slow-oscillation electrooculogram was then performed using the *Biomedica Mangoni BM6000-MAXI* device. The patient was pre-adapted for a period of 15 minutes.

Visual acuity was 20/20 in both eyes. Biomicroscopy and tonometry were normal. On fundus examination, the metallic yellow appearance was noted, with vessels of intense colour (Figure 1). On ERG, rod response (dim white light under scotopic conditions) was unrecordable (Figure 2-A). The maximum response (strong white light under scotopic conditions) showed a decrease in b-wave amplitude (Figure 2-B). The oscillatory potential was altered (Figure 2-C). Photopic response (Figure 2-D) and the response to the 30 Hz flicker (Figure 2-E) were within normal limits. Electrooculography (EOG) response was 208% in the right eye and 222% in the left eye (Figure 2-F). The Mizuo-Nakamura phenomenon was observed.

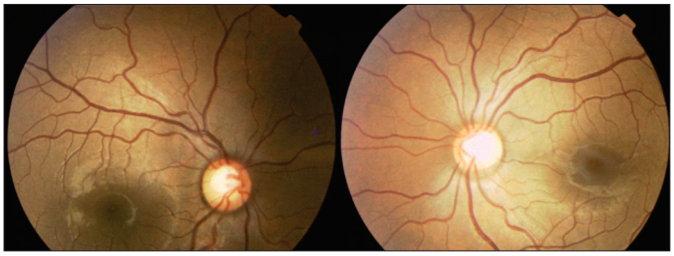

Figure 1. Retinography showing the metallic yellow appearance with vessels of intense colour, typical of Oguchi disease

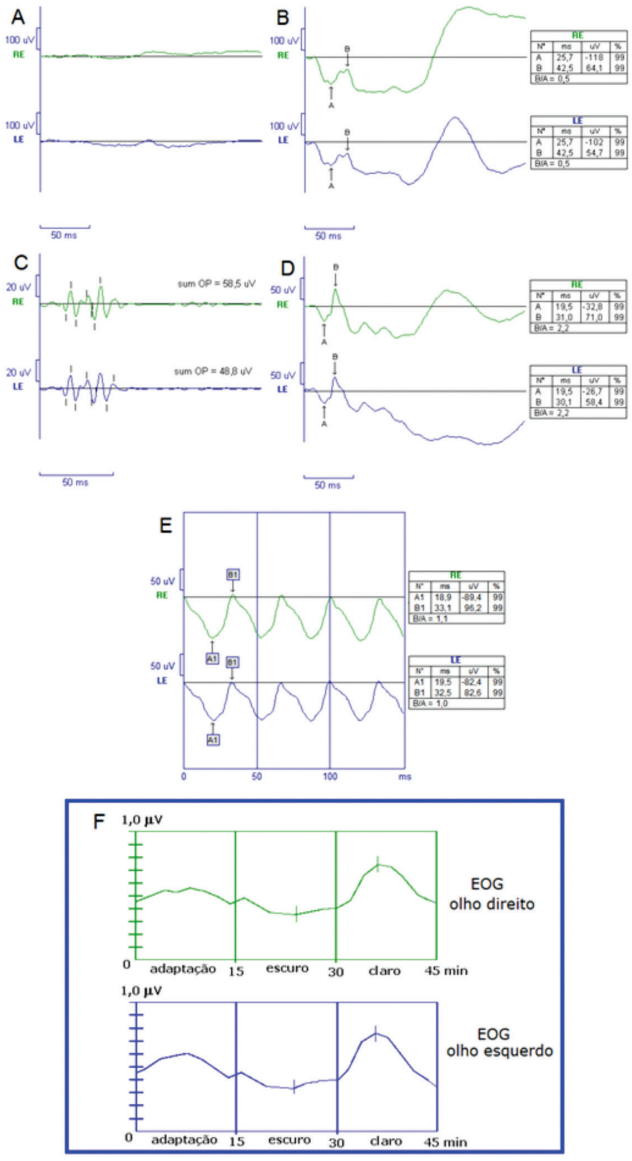


Figure 2. A) Rod response (dim white light under scotopic conditions) was unrecordable. B) The maximum response (strong white light under scotopic conditions) showed a decrease in b-wave amplitude (Figure 2-B). C) Oscillatory potential response. D) and E) The photopic response and the response to the 30 Hz flicker were within normal limits. F) EOG within normal limits in both eyes

## Discussion

Oguchi disease is a very rare form of SNB. Few cases have been described in Brazil[3]. Perhaps the most important feature of the electrical response in Oguchi disease is the lack of correspondence between visual sensitivity and b-wave amplitude; even though the scotopic curve tends to normalise after 4 hours of dark adaptation, b-wave amplitude remains very low [4].

On fundus examination, the typical metallic yellow appearance of Oguchi disease was noted.[5,6] The Mizuo-Nakamura phenomenon was present[2,5,6] and rod adaptation was markedly slow, as this was probably a case of Oguchi disease type I. It is important to note that the Mizuo-Nakamura phenomenon seen on fundus

examination may be unrelated to the dark adaptation curve[4].

On ERG, rod response (dim white light under scotopic conditions) was unrecordable and the maximum response (strong white light under scotopic conditions) showed a decrease in b-wave amplitude. This is consistent with the literature, which reports that under scotopic conditions, a-wave amplitude is normal and b-wave amplitude is markedly decreased or absent[7]. Responses within normal limits to the 30 Hz flicker confirm that the cones are fully functional, which is common in Oguchi disease[4].

The EOG within normal limits is in line with some previously-reported cases[4], but not with others[8]. Histological examination shows that the rods are present and that there is secondary adaptation, indicating that, under certain circumstances, these rods can be functional. It is also believed that the rods may have difficulty converting light energy into nerve impulses due to the lower concentration or absence of photosensitive pigments[9]. In any case, the normal EOG response confirms the hypothesis that the light-sensitive phase of the EOG may not depend only on rod function.

Electrofunctional tests are very important in the definitive diagnosis of Oguchi disease. In this case, the mismatch between EOG and ERG was clear. With regard to rod function, normal EOG responses contrast with the absence of rod response in the ERG under scotopic conditions. Further studies are needed to understand the complex electrofunctional mechanism of the disease and to better determine the origin of the light-sensitive components of the EOG.

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