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Case Report

A Rare Case of Oguchi Disease

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Abstract

Oguchi disease is a rare autosomal recessive disorder and the patients present with congenital stationary night blindness with slow dark adaptation. Two causative genes have been reported till date. The fundus shows typical golden sheen pattern which disappears when they remain in a darkened environment for few hours. We report a case of young girl who presented with non-progressive night blindness for 10 years and on examination typical fundus finding of Oguchi disease was revealed. The typical golden sheen fundus disappeared after 3 h of dark adaptation. This case is reported for its rarity as only <50 cases have been reported till date.

Key words: Dark adaptation, Mizuo-Nakamura, Oguchi

INTRODUCTION

Oguchi is an autosomal recessive disease characterized by congenital stationary night blindness and Mizuo-Nakamura phenomenon in which the fundus has a typical golden sheen pattern. It is a very rare condition and only 50 cases have been described in the literature till date.

CASE HISTORY

A 20-years-female presented to our OPD with complaints of defective night vision since 10 yrs. The patient was normal 10 years ago after which she noticed defective night vision which has not progressed since then. Day time vision is normal. There was no history of trauma, headache, field defects or any other significant positive complaints. No similar complaints in the family. She is single child to her parents. There was no history of consanguineous marriage.

On examination, anterior segment was normal in both the eyes. Vision in both eyes was 6/6 for distance and N6 for near vision. Color vision was normal. Fundus examination revealed normal optic disc and vessels with diffuse golden-yellow metallic sheen in the background retina [Figure 1a, 2a, 3a]. Oguchi was suspected and the patient was subjected to dark adaptation.

After prolonged dark adaptation of 3 h, the golden sheen disappeared with appearance of a normal reddish background retina [Figure 1b, 2b, 3b]. Field study was normal. Confirmation of Oguchi disease was made by electroretinogram (ERG) which showed absent A and b waves under light and increased wave in dark. The patient was explained about the condition and its prognosis. The patient was advised for regular follow-up.

DISCUSSION

Oguchi disease was first described by a Japanese Ophthalmologist Chuta Oguchi in the year 1907. Its characteristic fundus appearance was then described in the year 1913 by Mizuo. Oguchi disease is a rare autosomal recessive disorder which presents as non-progressive night blindness since childhood or birth with normal day vision. The typical fundus appearance is a diffuse or patchy, silver-gray, or golden-yellow metallic sheen with retinal vessels standing out in relief against the background. On prolonged exposure to dark adaptation for 3 h or more the unusual discoloration disappears with appearance of normal looking retina. This phenomenon is known as the Mizuo-Nakamura phenomena which is thought to be caused by the overstimulation of rod cells.[1]

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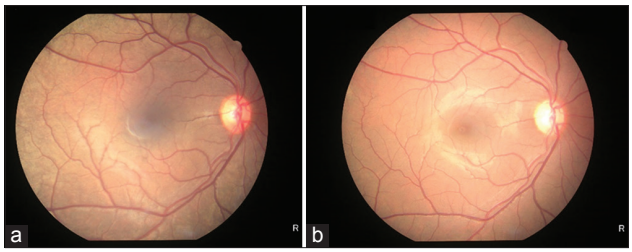

Figure 1: (a and b) Right eye fundus appearance before and after dark adaptation

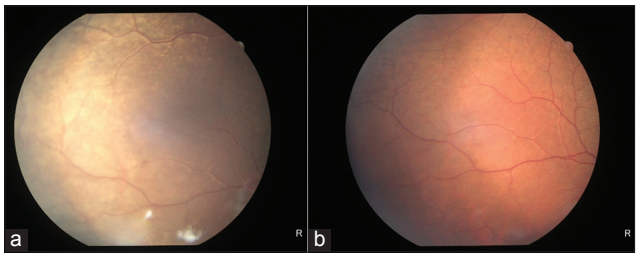

Figure 2: (a and b) Right eye fundus appearance before and after dark adaptation

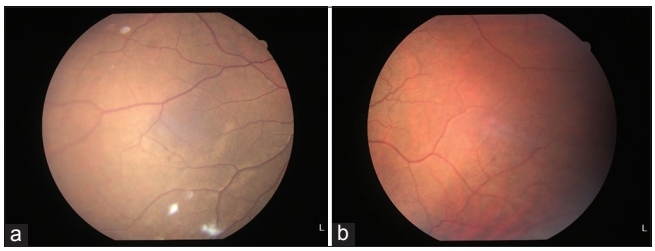

Figure 3: (a and b) Left eye fundus appearance before and after dark adaptation

Mutations in arrestin gene located on chromosome 2q37 or the rhodopsin kinase gene located on the chromosome 13q34 is found to be responsible for this disease. Rhodopsin kinase along with arrestin shuts off rhodopsin after it has been activated by a photon of light.

Yamamoto *et al.*[2] found that few cases of Oguchi are caused by defects in the GRK1 gene.

In ERG, the A- and b-waves on single flash are decreased or absent under lighted conditions and increased after prolonged dark adaptation. The b waves are nearly undetectable in the scotopic 0.01 ERG and nearly negative scotopic 3.0 ERGs.

Dark-adaptation studies have shown that rod thresholds are highly elevated decrease several hours later which results in recovery to the normal or nearly normal level.

Maw *et al.*[3] reported two Indian brothers with night blindness at an early age. 28-year-old brother presented with light-dependent golden fundus discoloration with 30-Hz flicker ERG responses. Under scotopic conditions, a white flash elicited a negative wave, whereas the response to a blue flash was extinguished. His 18-year-old brother had similar findings in his right eye. His fundus showed macular degeneration, mottled retinal pigment epithelium in the posterior pole and midperiphery, vitreous floaters, pale disk, and sheathed, attenuated vessels.

There is no specific treatment for the disease. Oguchi can lead to visual field defects at later stage. The patient should be on regular follow-up.

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