

CASE REPORTS

OGUCHI DISEASE – TWO PATIENTS WITH VARIABLE GENE MUTATION AND OPTICAL COHERENCE TOMOGRAPHY FINDINGS

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ABSTRACT

Oguchi disease is a rare form of congenital stationary night blindness which has autosomal recessive inheritance. It is characterized by typical clinical features in which there is golden yellow tapetal reflex over the fundus which disappears after prolonged dark adaptation. This clinical characteristic is known as "Mizuo-Nakamura phenomenon". Patients with Oguchi disease present with night blindness which is non-progressive and not associated with decreased vision, colour desaturation or visual field defects.

We examined two patients who presented with night blindness since childhood. A 4 year boy and a 6 year old girl when examined in detail revealed Mizuo Nakamura phenomenon on fundus examination. They were further investigated with electrophysiological tests, optical coherence tomography and genetic studies to make a diagnosis of Oguchi's disease.

Keywords: Congenital stationary night blindness, Mizuonakamura phenomenon, Oguchi disease.

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INTRODUCTION

Oguchi disease is a rare autosomal recessive form of congenital stationary night-blindness that is characterized by a golden tapetal fundus reflex. The color of the fundus reflex in the light adapted state has also been described as golden-yellow, gray-white, and yellow-white. This reflex can appear either homogeneous or in streaks in the fundus and disappears after prolonged dark adaptation^{1,2}.

This short communication presents two cases who reported with night blindness, when examined in detail came out to be cases of Oguchi's disease with different optical coherence tomography (OCT) findings. The aim to report these cases was to acquaint ophthalmologists about the rare presentation of congenital stationary night-blindness.

CASE REPORT

Case-1

A 4 year old boy presented with non-progressive poor vision at night for the last 2

years that was not associated with other ocular or systemic complaints. Visual acuity was 6/6 and posterior segment examination revealed an unusual golden yellow sheen all over fundus more marked in periphery outside the arcades with normal foveal reflex and no pigmentary changes, flecks, disc pallor or attenuation of vessels. Fundus photographs after 8 hours

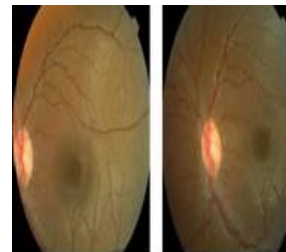


Figure-1: Fundus photograph before and after prolonged dark adaptation.

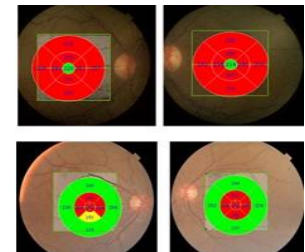


Figure-2: OCT case 1 and Case 2.

of patching revealed Mizuo Nakamura phenomenon which is characterized by loss of the golden sheen and tapetal reflex after prolonged dark adaptation (fig-1) OCT (Topcon 3D OCT-1 Maestro) showed normal central foveal thickness but reduced retinal thickness in parafoveal region (fig-2). Electro-retino-gram (ERG) of the patient showed grossly depressed a and b wave

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amplitudes. Genetic studies revealed SAG gene mutations.

Case-2

A 6 year old girl presented with poor vision at night for the last 4 year that was not associated with photophobia, redness, ocular trauma, glasses, cataract, glaucoma, or any ocular surgery. Visual acuity was 6/6, anterior segment examination was unremarkable while posterior segment examination revealed golden sheen over the fundus and Mizuo Nakamura phenomenon. OCT (Topcon 3D OCT-1 Maestro) showed reduced thickness in both foveal and para foveal area (fig-II). Scotopic ERG of patient revealed grossly depressed rod responses and genetic studies revealed GRK1 gene mutations.

Parents were counselled about the stationary nature of disease and advised to limit night time activities.

DISCUSSION

Oguchi disease is a very rare form of congenital stationary night blindness in which patients usually present with non progressive nyctalopia and Mizuo-Nakamura phenomenon.

The ERG may show either markedly reduced/absent rod response with normal cone function or affect both scotopic and photopic ERG. The two cases that we have reported here revealed markedly diminished rod responses with normal cone response on ERG.

Genetic studies have shown two causative mutant genes named as SAG (S antigen) and GRK1 (G protein coupled receptor kinase-1) for the pathogenesis of oguchi disease in Japanese and European population³. Some authors demonstrated GRK1 gene mutations as causative agent for oguchi disease in Pakistani population⁴.

We found SAG gene mutation in case-1 while GRK1 gene mutation in case-2.⁴

OCT (Topcon 3D OCT-1 Maestro) in Oguchi disease has shown changes in parafoveal region and some authors concluded the effect of prolonged dark adaptation on these changes with special focus on integrity of inner segment-outer segment (IS-OS) junction^{5,6}. However, we found thinning in parafoveal region with normal foveal thickness in case-1 and reduced thickness in both foveal and parafoveal region in case-2.

CONCLUSION

It is concluded that Oguchi disease is a rare entity, therefore, it is difficult to ascertain the exact relationship between various OCT pattern and genetic mutations.

CONFLICT OF INTEREST

This study has no conflict of interest to declare by any author.

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