DOI: 10.21276/aimdr.2020.6.5.OT2

Case Report ISSN (O):2395-2822; ISSN (P):2395-2814

Oguchi Disease or Stationary Congenital Night Blindness: A Case Report

Purva Modi[1], Bhavina Chaudhari[1], Vipul Prajapati[2], Anshul Jain[3]

[1]Third year Resident, M & J Western Regional Institute of Ophthalmology, Civil Hospital, Asarwa, Ahmedabad, India.
[2]Assistant Professor, M & J Western Regional Institute of Ophthalmology, Civil Hospital, Asarwa, Ahmedabad, India.
[3]Fourth year Resident, M & J Western Regional Institute of Ophthalmology, Civil Hospital, Asarwa, Ahmedabad, India.

Received: July 2020
Accepted: July 2020

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.

Name & Address of Corresponding Author
Dr Purva Modi,
Third year Resident,
M & J Western Regional Institute of Ophthalmology,
Civil Hospital, Asarwa,
Ahmedabad, India
E-mail: drprm169@gmail.com

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 dimished in both eyes. The cone ERG was normal in both eyes.

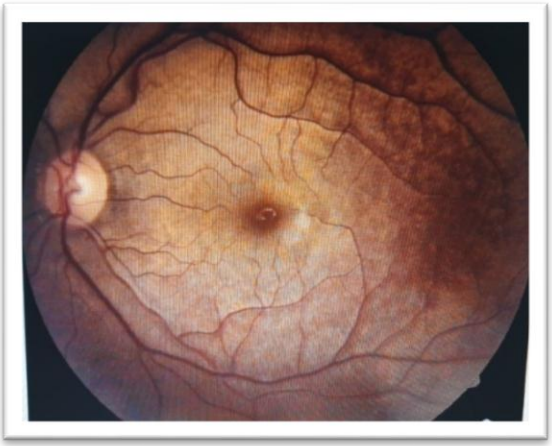

Figure 1: OS (Right eye)

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]

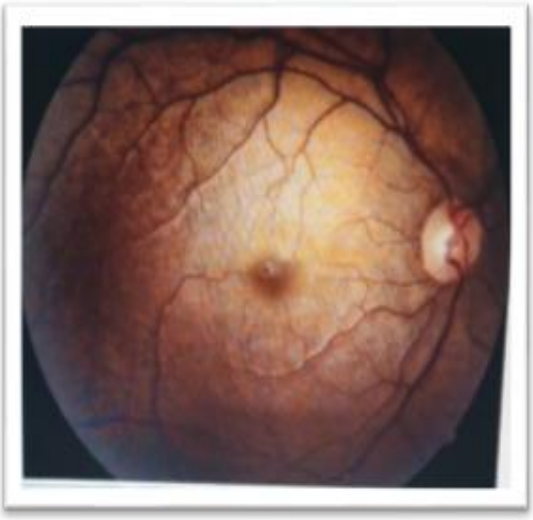

Figure 2: OS (Left eye)

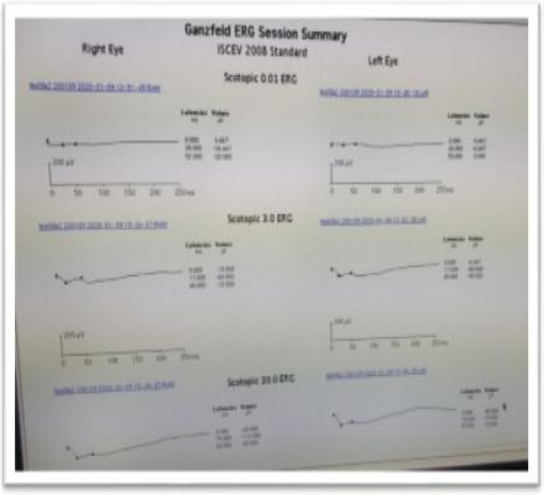

Figure 3: Electroretinogram

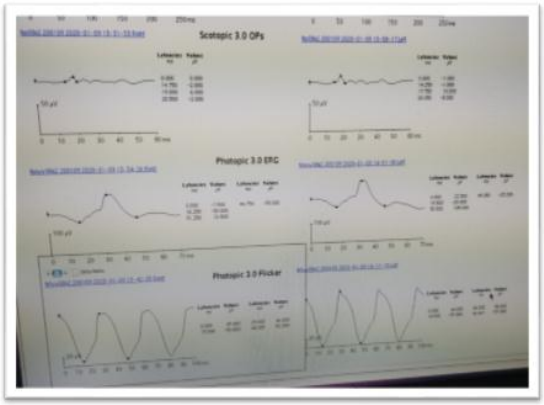

Figure 4: Electroretinogram

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

CONCLUSION

The diagnosis should be made when hemarlopia 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]

REFERENCES

1. Kuroda M, Hirami Y, Nishida A,et al. Nippon Ganka Gakkai Zasshi. A case of Oguchi Disease with Disappearance of Golden Tapetal-Like Fundus reflex After Vitreous Resection. 2011;115(10):916-923.
2. Agrawal R, Tripathy K, Bandyopadhyay G, Basu K. Mizuo-Nakamura phenomenon in an Indian male. Clin Case Rep. 2019;7:401-403. 10.1002/ccr3.1990.
3. Kalpana S, Muthayya M, Doctor PP. Oguchi disease. J Postgrad Med (serial online). 2006;52:143-4.
4. Yamamoto S, Hayashi M,Takeuchi S, Shirao Y, Kita K, Kawasaki K. Normal S cone electroretinogram b-wave in Oguchi's disease. Br J Ophthalmol. 1997;81 : 1043-5.
5. Maw M, Kumaramanickavel G, Kar B, John S, Bridges R, Denton M. Two Indian siblings with Oguchi disease are homozygous for an arrestin mutation encoding premature termination. Hum Mutat. 1998;1 ;S317-9.
6. Maw MA, John S, Jablonka S, Muller B, Kumaramanickavel G, Dehlmann R, et al. Oguchi disease : suggestion of linkage to markers on chromosome 2q. J Med Genet. 1995;32:396-8.

How to cite this article: Modi P, Chaudhari B, Prajapati V, Jain A. Oguchi Disease or Stationary Congenital Night Blindness: A Case Report. Ann. Int. Med. Den. Res. 2020; 6(5):OT04-OT06.

Source of Support: Nil, **Conflict of Interest:** None declared