Oguchi disease

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A 9-year-old healthy boy was brought by his mother for non-progressive defective vision in the evenings in both eyes of four years duration. His day-time vision was normal. He did not give history of trauma, previous ocular or systemic disease. His father has similar history of defective night vision. He was born of second-degree consanguinity and other two siblings were normal. In both eyes, uncorrected visual acuity was 6/6, N6. Slit lamp examination of anterior segment was normal. Color vision & field of vision (Octopus static white on white perimetry) were normal in both eyes. Fundus examination of both eyes revealed a patchy grayish metallic phosphorescent sheen. Optic disc, macula, retinal vessels were normal. When the boy was subjected to dark adaptation for 3 hours, the fundus background colour changed from gray to red. (Mizuo Nakamura phenomenon) [Figures 1-4].

The ERG response, schematically illustrated in Figure 5, consists of an initial negative wave, the a-wave, followed by a second positive wave, the b-wave. The a-wave is derived from the retinal photoreceptors: the rods and cones. It was originally thought that the Müller cells produced the b-wave, but more recent findings suggest that the b-wave is actually a result of bipolar cell activity. Since the nucleus of the Müller cell resides within the bipolar cell layer, this response is therefore indicative of changes occurring in that particular layer. There is no contribution to the ERG from the retinal ganglion 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 [Figure 6]. The Cone ERG was normal in both eyes [Figure 7].

Examination of the patient’s mother & other two siblings were unremarkable. His father could not be examined as he was a convict under custody.

Discussion

Oguchi’s 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 the dark environment; dark-adaptation study demonstrates that highly elevated rod thresholds decrease several hours later and eventually result in a recovery to the normal or nearly normal level. The fundus has a diffuse or patchy appearance, silver-gray or golden-yellow metallic sheen and the retinal vessels stand out in relief against the radiant background. A prolonged dark adaptation of three hours or more, leads to disappearance of the unusual discoloration of the normal reddish appearance, called Mizuo-Nakamura phenomenon.[1]

Oguchi’s disease is also unique in the electoretinographic responses in the light- and dark-adapted condition. B wave amplitude is reduced in rod response and cone response is normal.[2] Recent identification of the arrestin gene mutation[3,4] in chromosome 2q[5] in patients with Oguchi’s disease may account for the characteristic fundus and functional abnormality. As rhodopsin kinase works with arrestin in shutting off rhodopsin after it has been activated by a photon of light, it has been proposed that some cases of Oguchi disease are due to defects in rhodopsin kinase.[6]

In our patient, the classical Mizuo phenomenon was present and the fundus discoloration returned to normal with dark adaptation. The differential diagnosis includes Stargardts disease, female carrier of retinitis pigmentosa, juvenile retinoschisis, progressive cone dystrophy. All these conditions may have fundus changes but without the classical Mizuo phenomenon. In ERG both rod and cone abnormalities are found.

This rare disease is more commonly encountered in Japan and is presented in view of its novelty.
Figure 1: Fundus picture of right eye showing light adapted retina with gray discoloration of retina.

Figure 2: Fundus picture of right eye after dark adaptation showing normal red background

Figure 3: Fundus picture of left eye showing light adapted retina with gray discolouration of retina.

Figure 4: Fundus picture of left eye after dark adaptation showing normal red background

Figure 5: Schematic diagram of the basic ERG responses defined by standard.

Figure 6: ERG: Rod/max combined/OPs response

Figure 7: ERG: Single Flash cone/30 Hz

References