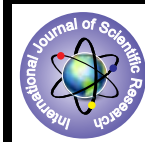


Utility of Optical Coherence Tomography in The Evaluation of Patient With Cherry-Red Spot



Medical Science

KEYWORDS : Cherry red spot, Optical coherence tomography

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ABSTRACT

A 4-year-old Japanese boy with 1-year history of unbalanced gait and epileptic seizure was referred to the clinic for an ophthalmological examination. Funduscopic examination revealed macular cherry-red spot. Optical coherence tomography (OCT) revealed increased reflectivity in the retinal ganglion cell layer in both maculae. We consider that it is important to note ophthalmic images using OCT are useful method for detecting neurological metabolic disorders presenting with a cherry-red spot in the macula.

INTRODUCTION

A macular cherry-red spot is a pathognomonic ocular manifestation of central retinal artery occlusion, traumatic retinal edema, and various forms of lysosomal storage diseases, such as GM1 gangliosidosis, GM2 gangliosidosis (Tay-Sachs disease, Sandhoff disease), Gaucher disease, Krabbe disease, Niemann-Pick disease, Farber disease, multiple sulfatase deficiency, metachromatic leukodystrophy, sialidosis, and galactosialidosis.^{1,2} The characteristic pale hue results from heavy deposition of lipid, sphingolipid, or oligosaccharide material in the ganglion cells of the retina at the macula, where these cells are several layers thick. In the center of the pale region lies the foveal pit which lacks ganglion cells, and thus continues to retain its reddish appearance.² Few reports have described the use of optical coherence tomography (OCT) to examine cases of cherry-red spot.³⁻⁶

We describe OCT findings in a 4-year-old boy with cherry-red spot.

CASE PRESENTATION

A 4-year-old Japanese boy with 1-year history of unbalanced gait and epileptic seizure was referred to the clinic for an ophthalmological examination. Visual acuity was not available. Slit-lamp examination showed no abnormalities in both eyes. Funduscopic examination revealed macular cherry-red spot (Fig. 1).

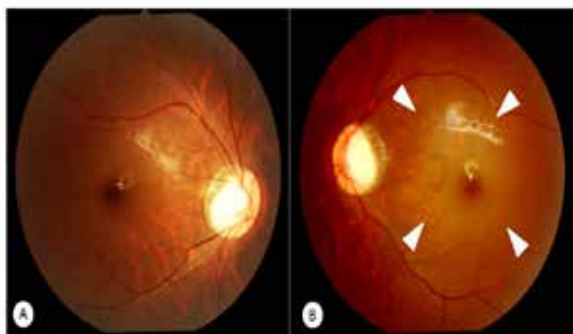


Fig 1. Funduscopic images of the right (A) and left (B) eyes. Cherry-red spot in the macula was more clearly detected in the left eye (arrows).

OCT revealed increased reflectivity in the retinal ganglion cell layer in both maculae (Fig. 2).

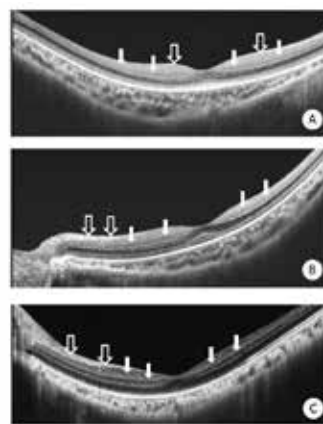


Fig 2. OCT images of a present patient (A and B) and a normal subject (C)

The boundary between the retinal nerve fiber layer (NFL; framed arrows) and the retinal ganglion cell layer (GCL; arrows) is clear in a normal subject (C). In contrast, the GCL appears hyperreflective in a patient (A and B).

Laboratory test results showed normal levels of β -galactosidase, hexosaminidase A and B. The patient was only followed-up by his pediatrician.

DISCUSSION

Few reports have described the use of OCT to examine cases of cherry-red spot.³⁻⁶ Rudich et al.³ reported three cases with Niemann-Pick disease. Yamazaki et al.⁴ reported a 16-year-old woman with galactosialidosis. Kersten et al.⁵ reported a 32-year-old woman with sialidosis. Zou et al.⁶ reported a 13-year-old boy with sialidosis. According to above reports, OCT revealed hyperreflectivity in the retinal ganglion cell layer. To the authors' knowledge, this is the youngest patient with cherry-red spot evaluated by OCT to be reported.

CONCLUSIONS

Although our findings were based on single case, we consider that it is important to note ophthalmic images using OCT are useful method for detecting neurological metabolic disorders presenting with a cherry-red spot in the macula. Furthermore, definitive characterization of cherry-red spot will require that additional genetic screening be undertaken in this present case.

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