Progressive Cone Dystrophy: a rare case report

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INTRODUCTION

Cone dystrophy (CD) is a term used to describe a group of eye disorders that affects the cone cells of the retina, either inherited or spontaneous appearance (1). Mutations have been described in about 12 genes and the three Mendelian inheritance patterns were found: autosomal dominant, autosomal recessive and X-linked. It can be divided into stationary and progressive disease. In the first one, symptoms arise at birth or in youth. On the other hand, in the second one symptoms progressively become worse over time and can cause completely blindness (2).

There is a huge variety of symptoms and the amount of vision loss is difficult to predict. As cone cells are involved with fine detailed vision, such as reading or recognizing faces, and with color perception, patients complains are related to that.

CD diagnose is clinically suggested by progressive visual acuity loss and inability of color discrimination, associated with day blindness and photophobia. Fundoscopy may show a bilateral symmetric macular atrophy. Diagnosis can be confirmed with electroretinogram (ERG) test (2).

METHODS

Medical record review.

RESULTS

We report the case of 57 years old male complaining of progressive visual acuity loss started 4 years before. Best visual acuity (BVA) was 20/400 in the right eye and 20/200 in the left eye. Fundoscopy showed a pale disc, decreased macular brightness and constricted arterioles. Electroretinogram showed a subnormal result in both eyes and nonrecordable photopic ERG.

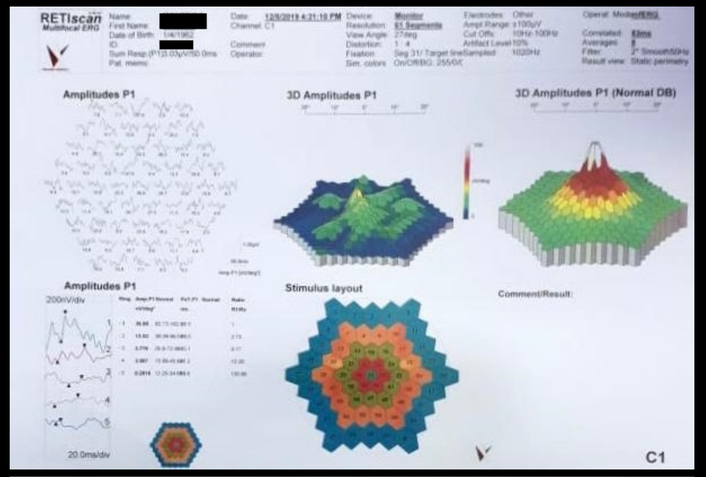
After diagnosed cone dystrophy disorder, the patient was instructed about his condition and adequate follow-up was provided.

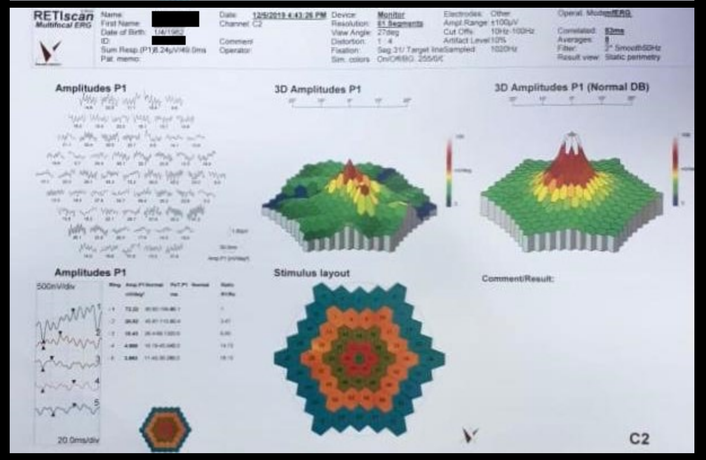
DISCUSSION

Besides clinical investigation, CD is diagnosed by ERG test, performed once in a bright room and once in a dark room. During that exam, retina is stimulated with flashing lights and through special contact lens-electrodes, it is possible to measure the electrical signals made by the cone and rod cells. A weak or absence signal indicates CD.

CD should not be confused with congenital color blindness, in which there is no retinal degeneration associated, only color deficits. There are a few differential diagnosis to be considered, such as cone-rod dystrophies, Leber's congenital amaurosis and Stargardt disease (3).

Up to now, there is no treatment or cure for CD. Treatment is directed to symptoms control, such as using tinted lenses or dark sunglasses in order to reduce photophobia and using magnifying devices to ensure better quality of life (1).





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