Case Report

A Case Presentation Emphasizing the Value of Full-field ERG in Retinal Dystrophies

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ABSTRACT: Purpose: A case presentation is used to discuss the value of full-field ERG in the diagnosis of cone-rod dystrophy. Case report: A 49 year old female patient known with amblyopia and myopic astigmatism since childhood, visited us complaining of decreased visual acuity and photophobia. Her BCVA was 0.6 in the RE and 0.4 in the LE. The fundus examination revealed normal colored and sharply limited papilla, normal macula and narrower vascularization. The Ishihara test was 10/15 in both eyes. The fundus autofluorescence and optical coherence tomography were normal. The DA 3 ERG was decreased and slightly delayed as well as the DA 0.01 ERG. Regarding the cone response, the LA 3.0 and 30 Hz flicker ERG were markedly reduced and delayed. The ERG findings indicated cone-rod dystrophy. At the 8 years follow-up examination, the BCVA decreased to 0.4 and 0.2 in the right and left eye, respectively, the Ishihara test was 1/15 in both eyes and the fundus examination showed a macula with no reflex, otherwise no changes compared to the first examination. The fundus autofluorescence and OCT were still normal. The ERG showed a much decreased and delayed rod response compared to the first examination, while the cone response was undetectable. Conclusion: Our case demonstrates the importance of standard full-field ERG in diagnosing CRD. The Full-field ERG responses were decreased and delayed, or even undetectable, while morphological examinations were still unchanged.

KEYWORDS: *ERG, normal clinical investigations, cone-rod distrophy*

Introduction

Cone rod dystrophies are a heterogenous group of hereditary, progressive retinal disorders that are characterized by the degeneration of photoreceptors [1]. Autosomal recessive, autosomal dominant and X-linked inheritance have all been reported [2]. The histopatological findings in these dystrophies showed cell death, decreasing outer segments of photoreceptors and abnormal photoreceptor synapses [3].

The principal and earliest symptom is bilateral, progressive visual loss [4]. Photophobia and colour vision abnormalities also occur early, while night blindness occurs later [1]. The fundus examination may show a wide range of aspects. Normal macula or fine macular lesions and pallor of the optic disc may be the only signs at early stage [1]. Retinal changes may also be similar to retinitis pigmentosa with bone spicule-like pigmentation [4]. White flecks similar to those seen in fundus flavimaculatus may be seen in the retinal periphery [2]. Attenuation of the retinal vessels, various degrees of retinal atrophy and even bull's eye maculopathy may be seen in patients with cone-rod dystrophies.

The diagnosis is based on the symptoms, retinal abnormalities seen at direct/indirect ophthalmoscopy, optical coherence tomography and fundus autofluorescence and changes in the standard electroretinography.

Standard full-field ERG is an objective method which emphasizes the function of the whole retina. It is also the key test in patients with cone-rod dystrophies, particularly at early stages [1]. ERG of CRD patients reveals reduced rod and cone ampitudes[2], with delayed a and b wave single flash photopic response and implicit time shift at the 30 Hz flicker response[1]. A trait of ERG findings in CRD is the early and predominant involvement of photopic over scotopic responses, unlike rod-cone dystrophies.

The purpose of this paper is to use a case presentation to emphasize the value of full-field electroretinoraphy in the diagnosis of cone-rod dystrophy.

Case Presentation

A female, aged 49 years old, known with amblyopia and myopic astigmatism (-4.00dsph, -3.00cyl/10° in the right eye and – 4.00dsph, -3.00cyl/180° in the left eye) visited the practice due to decreased visual acuity and markedly photophobia. The ophthalmological exploration gave a best corrected visual acuity (BCVA) of 0.6 in the right eye (RE) and 0.4 in the left eye (LE), Ishihara test 10/15 in both eyes, normal intraocular pressure and normal anterior pole at

biomicroscopy. The fundus examination revealed normal colored and sharply limited papilla, macula with a slightly decreased reflex and narrower vascularization (Fig.1) The fundus autofluorescence and optical coherence tomography were normal. The patient was sent for electrophysiological testing. The DA 3 ERG was decreased and slightly delayed as well as the DA 0.01 ERG. Regarding the cone response, the LA 3.0 and 30 Hz flicker ERG were markedly reduced and delayed. The clinical and ERG findings indicated cone-rod dystrophy (Fig.3).

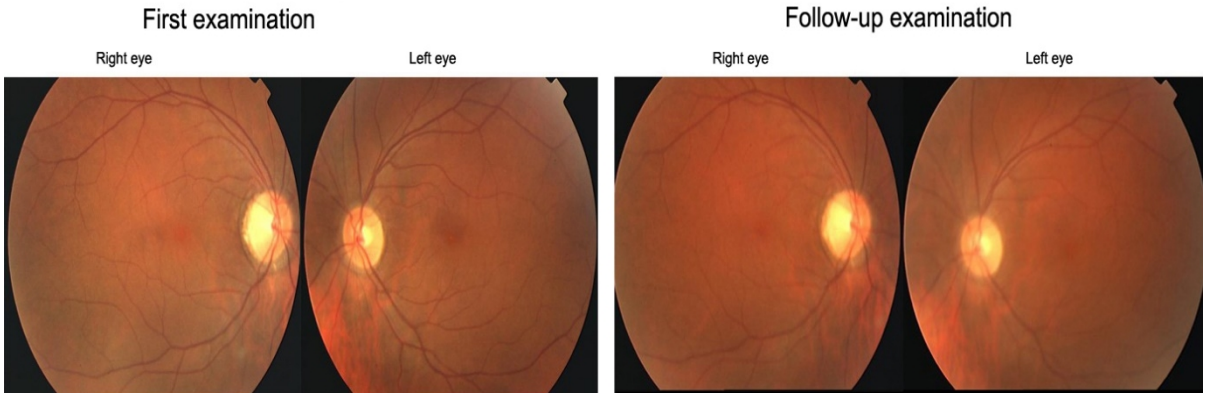

Fig.1. Fundus appearance at first and follow-up examination

At the 8 years follow-up examination, the patient complained of night visual problems, apart from photophobia and severely decreased BCVA (0.4 in the RE and 0.2 in the LE). The Ishihara test was 1/15 in both eyes and the fundus examination showed a macula with no reflex, otherwise no changes compared to the first examination (Fig.1). The fundus autofluorescence and OCT were still normal (Fig.2). The ERG showed a much decreased and delayed cone-rod response (DA 3 ERG) and rod response (DA 0.01 ERG), while the cone response was undetectable (LA 3 ERG and 30 Hz flicker) (Fig.3).

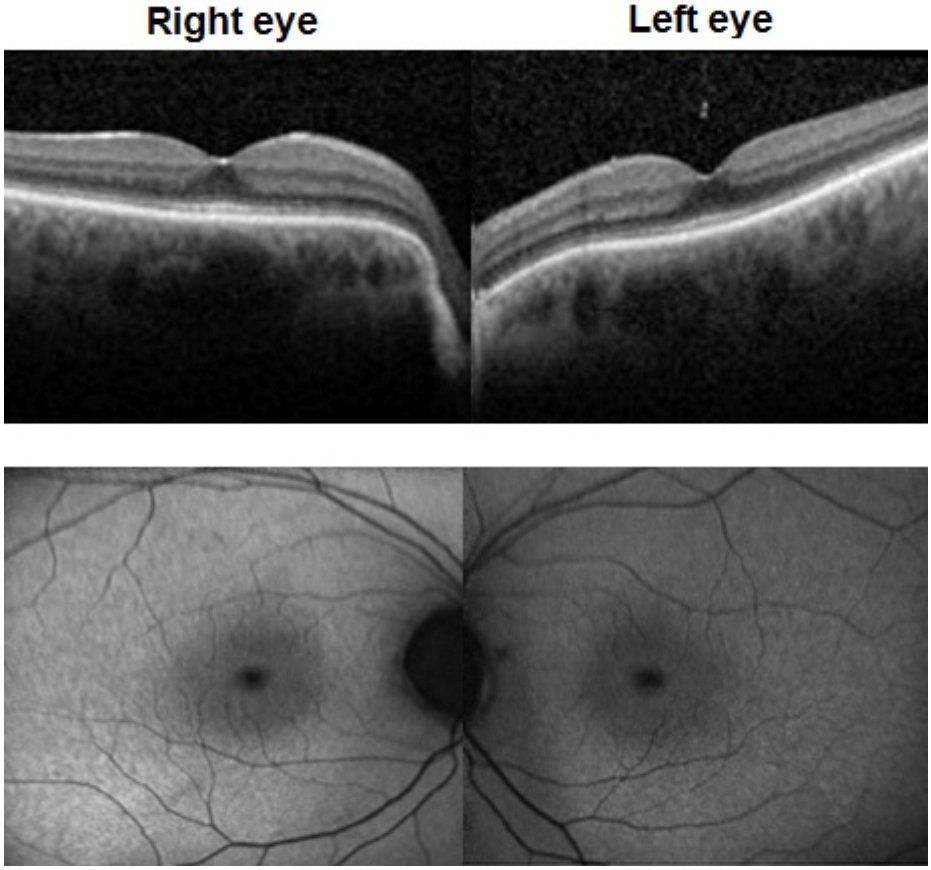

Fig.2. Normal OCT and fundus autofluorescence at the follow-up examination

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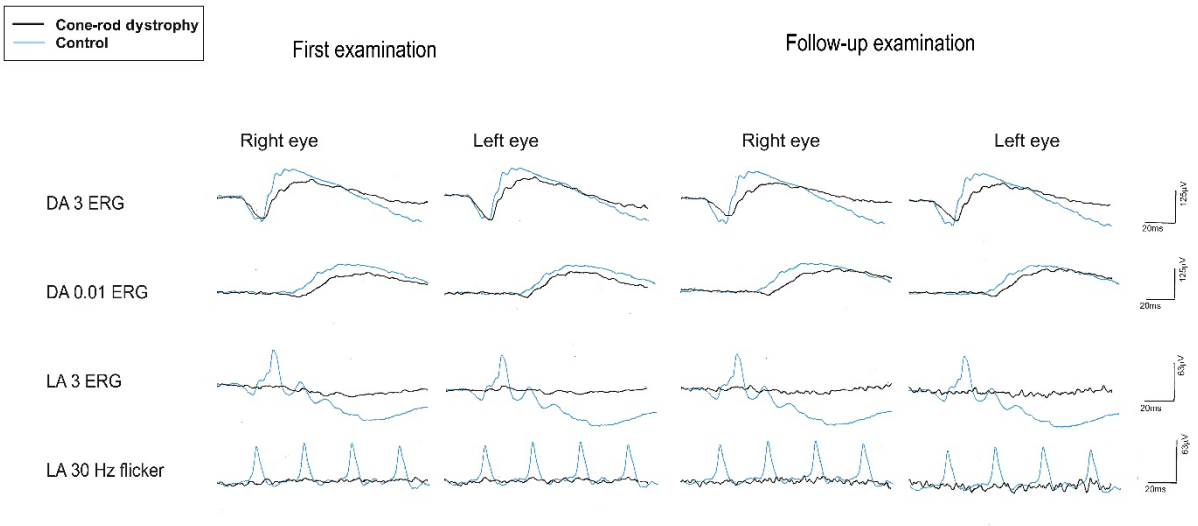

Fig.3. ERG findings in a CRD patient (black) compared to a normal control (blue)

Discussions

Cone-rod dystrophy has a prevalence of 1/40000 [1] and presents a wide range of clinical, morphological and functional aspects [5]. Usually, the age range for onset is between the first and third decades of life[4] with severe vision impairment in early adulthood. The symptoms may vary from mild to severely vision loss, photophobia, nystagmus, nyctalopia and colour vision abnormalities. The fundus may range from normal to bull's eye maculopathy.

In cases of patients with mild to moderate symptoms, absent or subtle changes in the fundus appearance and late onset an improper diagnosis can be made [5].

In our case, the patient presented with reduced visual acuity and a slightly decreased macular reflex. The morphological findings (optical coherence tomography and fundus autofluorescence) were within normal range. Due to the refractive error, the patient was diagnosed since childhood with amblyopia. The progression of vision loss and the appearance of photophobia required to carry out a full-field electroretinogram which emphasizes the function of the whole retina. The decreased rod response and the impaired cone function together with the clinical findings led to the diagnosis of cone-rod dystrophy.

The diagnostic was established on the basis of symptoms and full-field electroretinogram, which is the key test is cone-rod dystrophies diagnosis and allows the differentiation between inherited retinal diseases. The ERG reveals cone and rod function impairment, with cone system earlier and more affected than rod system.

Conclusion

Establishing the proper diagnosis in atypical and late onset cone-rod dystrophies may be difficult but the full-field electroretinogram is the most useful test in these cases, especially when the morphological investigations are within normal range.

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