

Congenital Cone-Rod Dystrophy to Progressing Central Vision Loss: A Low Vision Case Report

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Abstract—Cone-rod dystrophy is considered to be one of the rare ocular diseases that arises hereditarily. The nature of the disease can easily be mistaken with other ocular condition due to its unusual characteristics. This case report covers extensive ocular examinations and proper management of the condition.

Keywords: cone-rod dystrophy, hereditary, extensive ocular examinations

I. INTRODUCTION

Cone-rod dystrophy is said to be one over 40,000 individuals; providing there are several types. One of which is autosomal recessive pattern, where both copies of genes in each cell have mutations. Both parents of the affected individual have a copy of mutated gene but they do not exhibit signs and symptoms of the condition. ^[1]Anatomically, cone receptors are located at the fovea which are responsible for perceiving central vision. Cone-rod dystrophy affects primarily the photopic system which contributes the highest visual acuity and colour vision.

In contrast with a common retinitis pigmentosa, also called as rod-cone dystrophy, resulting to primary loss of rod photoreceptors and further progresses to cone receptors, CRDs imitates the opposed sequence of events. It is characterized by primary loss of cone receptors that progresses to the involvement of rod receptors. Loss of central vision, visual acuity, photoaversion and colour vision anomalies are the primary symptoms of CRD that can later progressto peripheral vision loss and night blindness upon rod degeneration. Cone-rod dystrophy is less prevalent but more severe and rapid than that of retinitis pigmentosa leading to earlier legal blindness. ^[2]

II. METHODS

A. Procedure

A 34-year old male, fair-skinned, bus service crew, presented for a comprehensive eye exam. Patient's chief complaint is blurring of vision at all distances with associated tearing for both eyes due to photophobia. The patient mentioned that he first experienced the symptoms in his early teenage years but was unable to seek propermedical attention because of financial problem and lack of facility in their area. The patient is a smoker and an occasional drinker.

His uncorrected visual acuity is 20/400 at 2m with a near acuity of J9 not readable for both eyes. Upon examining the patient while reading the chart, it is noticeable that the patient cannot fixate centrally. He is either looking inferiorly or laterally. Introduction of pinhole does not improved the patient's visual acuity which implies the presence of ocular disease. Motility test reveals jerky movements in version and duction but saccadic movement test was unable to perform because the patient is having a hard time locating the targets.

External examination test was performed and reveals no significant findings. The cornea and crystalline lens are transparent and has no opacities. Van Herick's angle of estimation is VH4. Keratometry findings shows irregular mires and yields an amount of -1.50Dcyl x 15 on OD and -1.00Dcyl x 15 on OS of external corneal astigmatism. Due to the inability

of the patient to fixate centrally, retinoscopy and ophthalmoscopy was unable to perform. Automated refraction results are $-2.00\text{Dsph} = -1.25\text{Dcyl} \times 20$ on OD and $-1.75\text{Dsph} = -0.75\text{Dcyl} \times 5$ on OS. The patient's visual acuity was slightly improved by ophthalmic lenses upon performing subjective refraction (20/200 at 2m). Farnsworth D-15 color vision test was unable to perform because the patient's colour vision is severely affected due to dystrophy.

B. Data Analysis

Consultation to an Ophthalmologist:

For further evaluation of his ocular condition, he was referred to a retinal specialist. The patient was asked regarding his ocular and medical history and was instilled with Tropicamide – a cycloplegic agent that aims to dilate the pupil before examination with a slit lamp. The doctor recommended fluorescein angiogram with auto fluorescence for further assessment of the condition.

The differential diagnoses included in this case:

Stargardt disease

Macular retinal dystrophy

Cone-rod dystrophy

- Stargardt disease is also called Stargardt macular dystrophy, juvenile macular degeneration, or fundus flavimaculatus. Vision loss is due to abnormal accumulation of a fatty yellow pigment (lipofuscin) in the cells within the macula.^[7]
- Macular retinal dystrophy is a rare genetic eye disorder that causes vision loss. It's caused by a pigment that builds up in the macula's cells. Over time, this substance can damage cells that play a key role in clear central vision.^[8]
- Cone-rod dystrophies (CORD) are inherited retinal degenerations characterized by cone degeneration which precedes the rod degeneration. Prevalence of CORD is estimated to be 1 in 40,000. The early manifestations of CORD include decreased visual acuity, color vision defects, and photophobia, with onset usually occurring in late childhood or early adult life, followed by progressive loss in peripheral vision and night blindness.^[2]

The final diagnosis is Cone-rod dystrophy for both eyes due to ocular signs and symptoms exhibited by the patient such as: decreased visual acuity, color vision defects, eccentric fixation, hypopigmentation of macula with clumps of retinal pigmented epithelium and dystrophy of cone receptors that progresses to the rods.

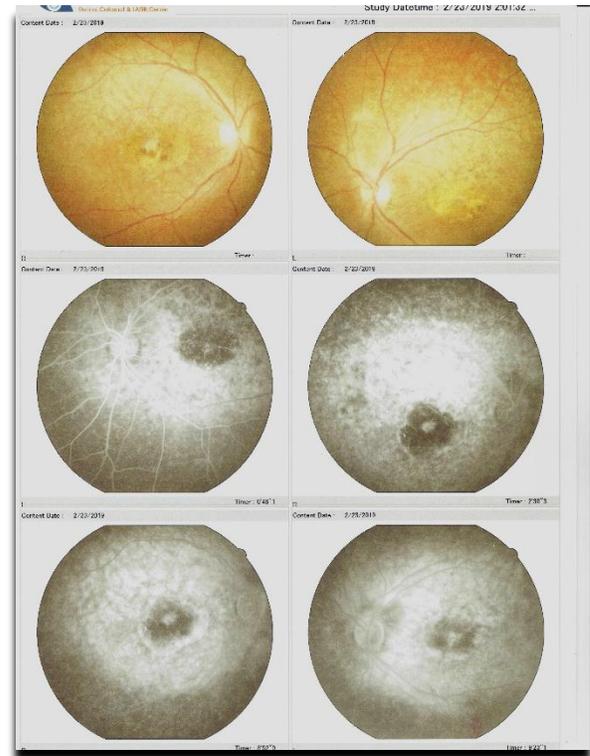


Figure 1. Initial retinal image and the accumulation of dye

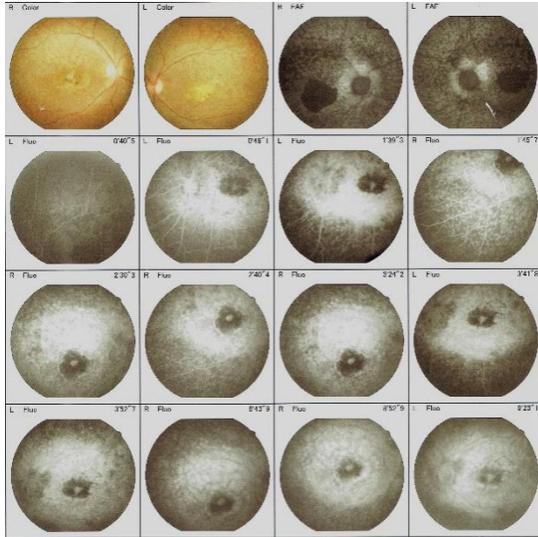


Figure 2. Accumulation of dye in the retina

III. RESULTS

Based on the results gathered: the initial eccentric fixation, color vision defects, hypopigmentation of macula with clumps of retinal pigmented epithelium and dystrophy that starts mainly at the cones radiating to the rods, the patient was diagnosed with Cone-rod Dystrophy.

IV. DISCUSSION

Collection of demographic data, ocular and medical history are necessary for a successful diagnosis and management of the patient. Blurring of vision at all distances is the chief complaint of the patient with an associated complaint of tearing due to photophobia. He then mentioned that he started to experience early symptoms in his teenage years but was unable to seek proper medical attention because of financial problem and lack of facility in their area. Preliminary tests, external and internal examinations were collected. Subjective refraction was also performed. Social history shows that he's a smoker and an occasional drinker. He was then referred to a retinal specialist and was recommended to have fluorescein angiogram with auto fluorescence that revealed the severity of the hypopigmentation of macula with clumps of retinal pigmented epithelium and the loss of foveal contour and was diagnosed with Cone-Rod Dystrophy

The patient was educated about the severity of his ocular condition. He was instructed to avoid direct sunlight and to cut his smoking habit. The patient was prescribed with $-1.75Dsph = -1.00Dcyl \times 180$ on both eyes tinted with yellow-amber shade to enhance the patient's contrast sensitivity and lutein – a lutein supplement for the preservation of the remaining cone receptors. The patient undergone low vision rehabilitation exercises such as: meal management, proper orientation when walking, sighted guide, and money management. This training was performed to help the patient to deal with his everyday routine.

V. CONCLUSION

Failure to consult medical assistance earlier led to rapid progression of central vision loss. Due to which, the progression of dystrophy was quick. There are no treatment for Cone-Rod Dystrophy yet, but early detection can slow down the progression. Low vision rehabilitation aims to improve the quality of life of an individual by helping them to easily perform their tasks independently. By enhancing patient's contrast, together with low vision training, the patient's performance was improved.

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