

## Hereditary Progressive Cone Dystrophy

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**Cone dystrophy is a rare hereditary eye disease involving retinal cone photoreceptors, which affects the central vision and color vision. Most patients present with defective color vision and significant photophobia.**

**We present two rare cases of progressive cone dystrophy who presented to the Ophthalmology Clinic and were treated conservatively.**

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Cone dystrophy is a heterogeneous rare disorder of retinal cone cells that manifest as a reduction in central visual acuity, defective color vision and photophobia<sup>1</sup>. The worldwide prevalence is around 1 in 40,000<sup>2</sup>. According to the clinical progression, cone dystrophy could be classified into two categories, stationary and progressive<sup>3</sup>. Stationary Cone dystrophy usually presents at birth or early childhood and nearly in all patients; the symptoms remain stable, unlike progressive cone dystrophy, in which the symptoms deteriorate over time<sup>4</sup>. Clinical assessment of the progression of the disease is an important aspect of managing patients with cone dystrophy as the pathogenesis of cone dystrophy is not clearly understood.

The aim of this report is to present two rare cases of progressive cone dystrophy and their management.

### THE CASE

The first patient was a 40-year-old Egyptian male, not known to have any medical illness, presented to with visual impairment since childhood, photophobia and difficulty to see in bright light with poor recognition of colors. The condition became worse during adulthood. He also complained of a small central black spot in his vision. There was no family history of any ocular diseases and there was no history of consanguinity.

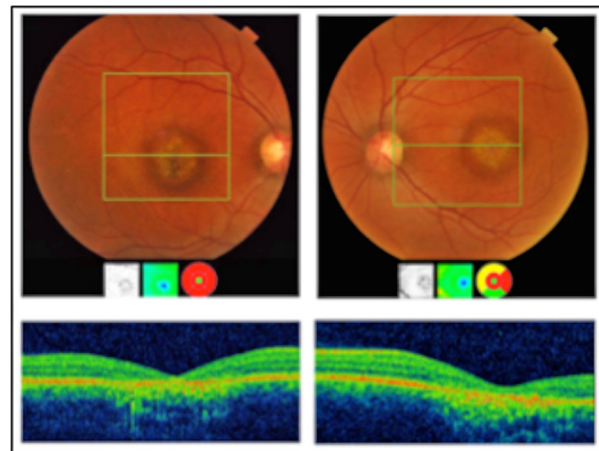
The patient's best-corrected visual acuity was 6/24 in both eyes. Ishihara Color Plates Test showed red-green color blindness. Anterior segment examination was normal in both eyes. Intraocular pressure was 15 mmHg in both eyes. Dilated Fundus Examination showed bilateral central macular atrophic lesions involving the fovea measuring two-disc diameter. Optic discs were healthy bilaterally.

Optical Coherence Tomography showed diagnostic pictures of cone dystrophy. The central macular thickness was 135  $\mu\text{m}$  OD and 150  $\mu\text{m}$  OS. Visual Field Test revealed paracentral scotoma in both eyes. Electroretinogram revealed reduced photopic response, although rod response was normal.

The second patient was a 28-year-old Indian female, not known to have any medical illness, presented with a history of visual impairment since childhood which deteriorated over the last year. The elder sister suffered from the same disease with positive consanguinity history as her parents are first degree cousins.

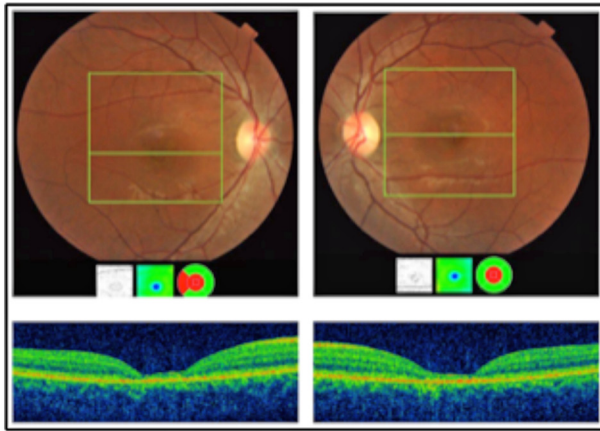
Her best corrected visual acuity was 6/18 in both eyes. Absolute red-green color blindness was detected on Ishihara color plates. Anterior segment examination was normal in both eyes. Intraocular pressure was 14 mmHg in both eyes. Dilated Fundus Examination showed a gross macular thinning with bilateral healthy optic nerves.

Optical Coherence Tomography showed foveal thinning with central macular thickness 170  $\mu\text{m}$  OD and 76  $\mu\text{m}$  OS, see figure 1. A visual field test showed central scotoma in both eyes, see figure 2. Significantly reduced cone response in Electroretinogram and preserved rod function was found.



**Figure 1: Optical Coherence Tomography of Patient I showing Bilateral Macular Thinning with Central Macular Thickness 135  $\mu\text{m}$  OD and 150  $\mu\text{m}$  OS**

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**Figure 2: Optical Coherence Tomography of Patient II showing Bilateral Macular Thinning with Central Macular Thickness 170 µm OD and 76 µm OS**

## DISCUSSION

Progressive cone dystrophies are a group of inherited disorders characterized by malfunction of the cone photoreceptors, occasionally extending to their post-receptor tracts. Major clinical features are a gradual deterioration of day light visual acuity with defective color vision and photophobia. Electroretinogram is used in diagnosis in which it records a subnormal or absent photopic cone response<sup>1</sup>.

The severity of symptoms in cone dystrophy is variable among affected patients. The degree of visual deterioration and its progression is unpredictable<sup>1</sup>.

Progressive cone dystrophy is characterized by deterioration of central vision with age. In many patients, visual acuity goes below 6/60, where they are labeled as legally blind. Peripheral vision is usually preserved; therefore, the patient could see well in dim illumination or at night, this is due to sparing of rod cells. However, in some cases, rod cells may be involved<sup>2,4</sup>.

Cone dystrophy has variable inheritance patterns. Cone dystrophies form a heterogeneous group of diseases genotypically and phenotypically<sup>5</sup>. Many cases of cone dystrophy are sporadic, while others could be inherited as an autosomal dominant<sup>6</sup>. Stationary Cone dystrophy is usually inherited as autosomal recessive while progressive cone dystrophy is usually inherited as autosomal dominant. Some cases could be inherited as X-linked recessive. Mutation in any of several genes play a role in inherited cone dystrophy; the most common genes are GUCY2D, CRX, ABCA4 and RPGR<sup>3,7</sup>.

The exact, underlying mechanisms that cause cone dystrophy are not fully understood<sup>2</sup>. Cone dystrophy is also found as a part of some syndromes, such as Alstrom syndrome which is due to the involvement of ALMS1 gene<sup>8</sup>.

Treatment is usually supportive in the form of dark glasses and low vision aids. Topical lubricants and miotics are used to relieve the photophobia<sup>9</sup>.

## CONCLUSION

**Progressive cone dystrophy is usually seen in association with some rod dystrophy. The two cases which we are**

**presenting are rare forms of pure cone dystrophy. Three major criteria used to diagnose these cases as pure cone dystrophy are reduced day light visual acuity, central field defect and intact night vision, ERG—showing reduced photopic response with intact rod response and defective color vision.**

**As of today, there is no cure for cone dystrophy and the treatment is largely supportive to relieve the symptoms; both patients were advised to use lubricating eye drops and tinted glasses to relieve photophobia and to maintain a substantial day light vision. They were also advised low vision aids.**

**The pathogenesis of cone dystrophy has not been clearly defined. It requires a multidisciplinary approach to assess its pathophysiology, which would involve molecular genetics, electrophysiological studies and psychophysics. Clinical assessment of the progression of the disease is an important aspect of managing patients with cone dystrophy.**

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