Case Report

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Retinopathy Associated with Cone Dystrophy

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Abstract

Progressive cone dystrophies represent a heterogeneous group of diseases with onset in later teens and adult age diagnosed by an abnormal photopic electroretinographic response and a normal or near-normal rod isolated response. We report the case of a young male with typical characteristics of cone dystrophy and a brief description about the inheritance pattern and pathology of the disease.Progressive cone dystrophies represent a heterogeneous group of diseases with onset in later teens and adult age diagnosed by an abnormal photopic electroretinographic response and a normal or near-normal rod isolated response. We report the case of a young male with typical characteristics of cone dystrophy and a brief description about the inheritance pattern and pathology of the disease.

Introduction

A cone dystrophy is a hereditary ocular disorder characterized by the loss of cone cells, the photoreceptors responsible for both central and colour vision [1, 2]. The most common symptoms of vision loss are: (age-range, from the late teens to the sixties), sensitivity to bright lights, and poor colour vision. Fluorescein angiography (FA) and Optical coherence tomography are useful adjuncts in the workup of someone suspected to have cone dystrophy, as it may detect early changes in the retina that are too subtle to be seen by ophthalmoscope. This pictorial shows the typical FA and OCT findings in a patient who was diagnosed to have cone dystrophy.

Discussion

This 48 year old male presented with complaints of gradual decrease in vision of four years duration. His BCVA was 20/1200 OD and 20/600 OS with a near vision of N36 OU. Colour vision was abnormal. Colour fundus photo showed the presence of a central hyperpigmented area with surrounding concentric areas of hypo and hyper pigmentation. Fluorescein angiography showed a corresponding area of central hypo-fluorescence with surrounding concentric areas of hyper, hypo and hyper-fluorescence resembling a 'target' sign. Optical coherence tomography revealed

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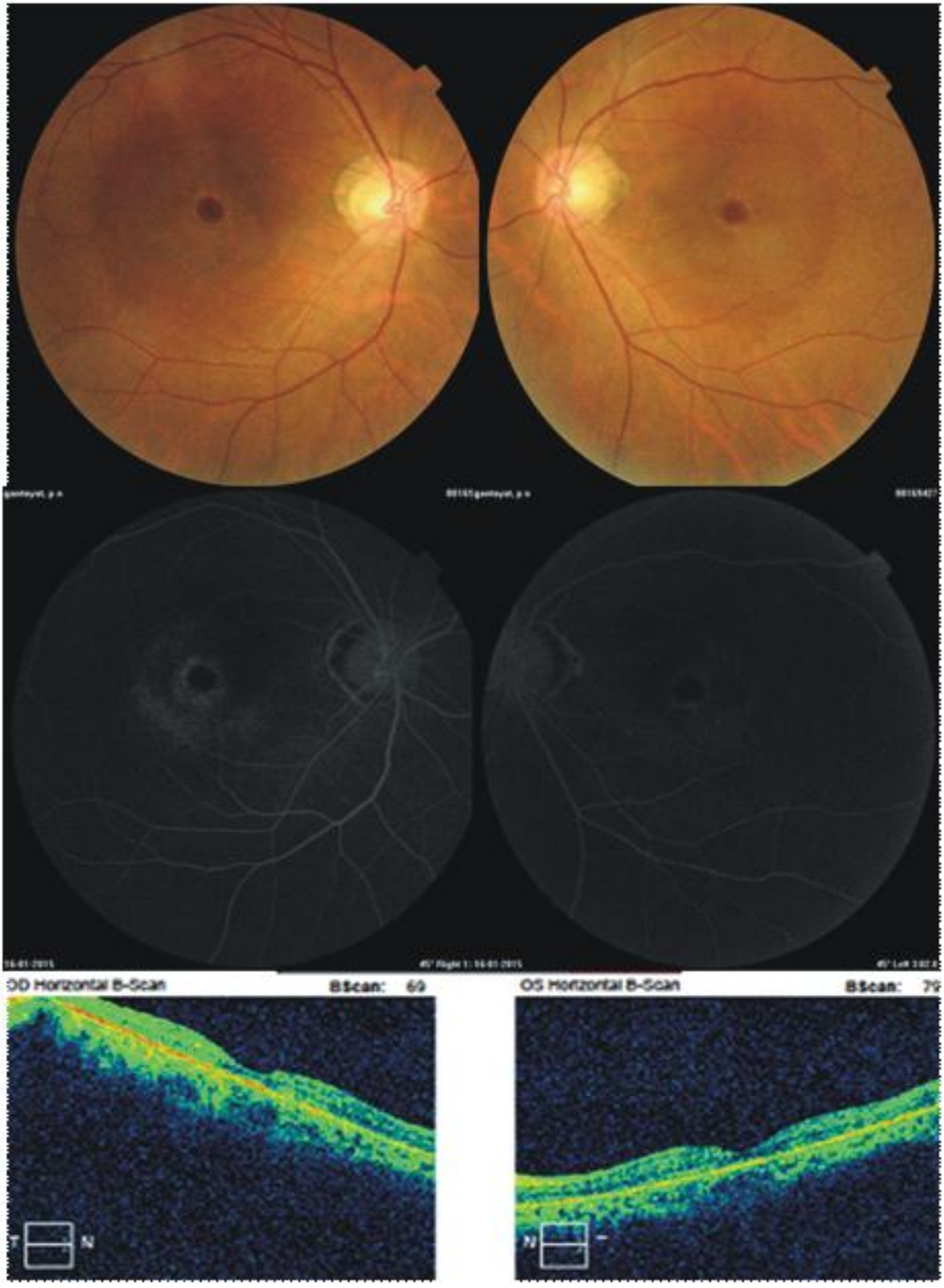
Fig. 1: This composite picture shows the colour fundus photos (upper tier) showing bulls eye maculopathy, FFA findings (middle tier) showing target appearance, OCT findings (lower tier) showing foveal atrophy in both eyes.



disruption of the IS-OS junction and foveal atrophy. ERG confirmed the diagnosis of cone dystrophy. The most common type of macular lesion seen during ophthalmoscopic examination has a bull's-eye appearance and consists of a doughnut-like zone of atrophic pigment epithelium surrounding a central darker area. In another, less frequent form of cone dystrophy there is rather diffuse atrophy of the posterior pole with spotty pigment clumping in the macular area. Rarely, atrophy of the choriocapillaris and larger choroidal vessels is seen in patients at an early stage. Visual field testing in cone dystrophy usually reveals a central scotoma. Photopic Electro-retinography (ERG) shows reduced single-flash and flicker response and a normal scotopic ERG. At least one type of autosomal dominant cone-rod dystrophy is caused by mutations in the guanylate cyclase 2D gene (GUCY2D) on chromosome 17. Though there is no cure for Cone dystrophy, certain supplements delay the progression of the disease. The beta-carotenoids, lutein and zeaxanthin, have been evidenced to reduce the risk of developing age related macular degeneration (AMD) [3], and may therefore provide similar benefits to Cone dystrophy sufferers. Consuming omega-3 fatty acids (docosahexaenoic acid and eicosapentaenoic acid) has been correlated with a reduced progression of early AMD, and in conjunction with low glycemic index foods, with reduced progression of advanced AMD,[4] and may therefore delay the progression of cone dystrophy.

References

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