Case Study: Heredomacular Degeneration

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Introduction

Heredomacular degenerations or macular dystrophies refer to a group of diseases which present with certain common features. These include an insidious onset with gradual progression, heredofamilial nature, bilaterality (may be assymetric) and occasional involvement of the central nervous system.

The typical lesion at the macula is described to have a pigmented and worm eaten appearance. It is typically clearly demarcated and is usually one disc diameter in size. There is an abnormal foveal reflex with a varied degree of associated vision loss.

The diagnosis is often made on clinical fundus examination though certain investigations such as multifocal electroretinography, electrooculography and optical coherence tomography may help in confirmation.

History

A 38-year-old male patient presented with progressive loss of vision over the past few years. The patient had history of reduced vision in daylight with a specific loss in the central field. The patient had no known precipitating factor and no family history of similar symptoms. The patient had been prescribed glasses in the past but they did not provide much improvement in vision.

Examination

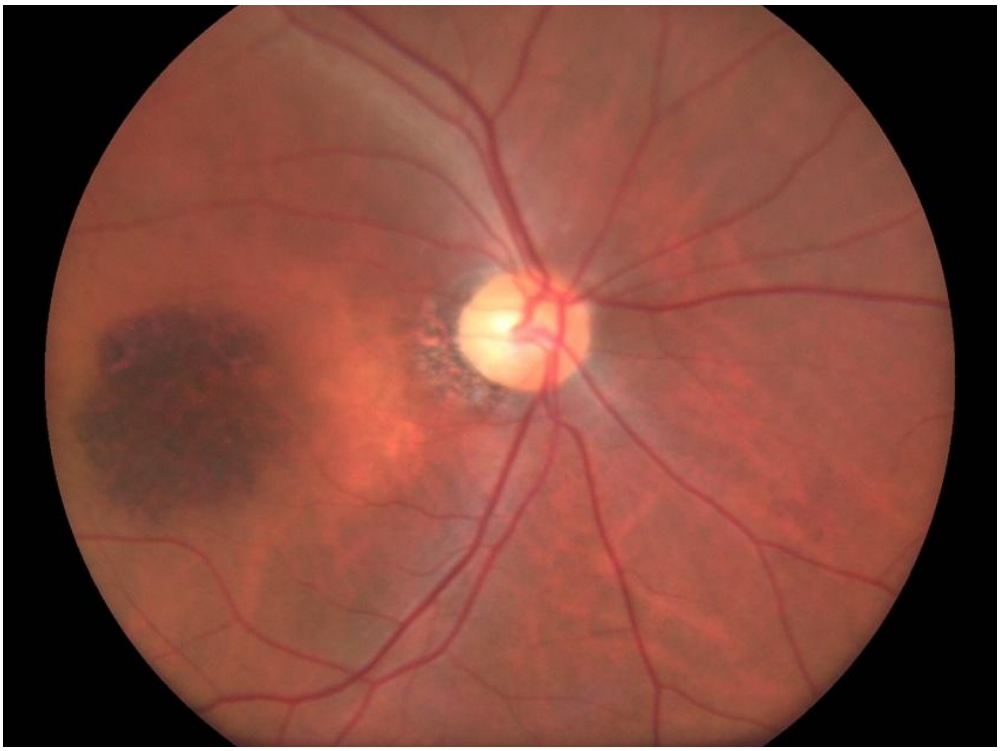
On examination, the visual acuity in both eyes was 20/120 with no further improvement with refraction. Anterior segment examination appeared normal and the pupillary reactions were brisk. Fundus examination revealed a well demarcated hyperpigmented lesion in the macula of both eyes with around 2 disc diameters in size and having a worm eaten appearance. (See fundus photographs).

Fundus photographs were taken with Optomed Smartscope PRO for keeping a record and following up progression of the disease.

Conclusion

The patient was diagnosed to have adult onset heredomacular degeneration and explained that the visual prognosis is guarded. In view of no definite treatment for this condition, the patient was prescribed low vision aids. Heredomacular degenerations tend to be progressive and a long-term follow-up is required.

Fundus photograph of the right eye showing a hyperpigmented lesion of the macula about 2 disc diameters in size. Also note the mild peripapillary atrophy around the disc in the temporal region.



Fundus image of the left eye showing a deeply pigmented lesion of about 2 disc diameters in the macula along with generalized chorioretinal degeneration. Notice the moth eaten appearance of the macular lesion.