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Case Report

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The Curious Case of Night Blindness: A Rare Case Report

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ABSTRACT

Introduction: Gyrate Atrophy of the choroid and the retina is a rare autosomal recessive disorder caused by a mutation in ornithine aminotransferase.

Case Report: We present an interesting case of a 55-year-old male who presented to us with complaints of night blindness, increase in number of minus glasses, senile cataract in both eyes, constriction of peripheral visual field and chorioretinal atrophic lesions in both eyes. Blood investigation revealed raised plasma ornithine levels at 690 micromole/L (normal range 30-90 micromole/L). There is progressive night blindness and visual field constriction due to progressive chorioretinal degeneration. It has now progressed to diminution of central visual acuity due to progressive macular and glaucomatous changes and cataract formation.

Conclusion: There is history of blindness in two out of 6 more siblings which indicates a hereditary pattern.

Key Words: Gyrate Atrophy, *Fundus Albipunctatus*, glaucoma, OAT gene, Ornithine amino transferase, Chorioretinal degeneration

INTRODUCTION

Gyrate Atrophyis a rare genetic disorder which leads to progressive chorioretinal degeneration, early cataract formation and myopia. It occurs due to mutations in the OAT gene, found on chromosome 10q26. Its inheritance pattern is mainly autosomal recessive.[1] Gyrate atrophy is caused by a deficiency in the enzyme ornithine aminotransferase (OAT), which results in a 10 to 20folds increase in plasma ornithine concentrations.[2,3] The most common pattern is the progression of atrophic lesions from the periphery towards the posterior pole.[4,5] Macular involvement occurs late in the disease. Supplementation of co-factor pyridoxine that partially takes over the function of missing enzyme, has been found to be helpful in few genetic variants of this disease.[6,7].

*Fundus Albipunctatus*is a rare autosomal recessive retinal disorder caused by mutations in RDH5 gene. It leads to deficiency of11-cis-retinol dehydrogenase 5 enzyme which plays integral role in visual cycle. [8,9] It is characterized by nyctalopia and the presence of whitish-yellow flecks in the retina. It involves mid periphery most of the time with variable number of flecks which get smaller or fade with age in some affected individuals, although night vision does not improve. [10]

While *Fundus Albipunctatus* typically does not worsen (progress) over time, some individuals may develop other eye conditions, such as macular degeneration, with the loss of cones, which can affect vision in bright light.

CASE REPORT

Our patient is a 55-year-old male presented to our retina clinic with progressive nyctalopia for 2 years which was gradual, painless and progressive in nature. He also developed diminution of vision in day light in both eyes for last 6 months. The visual acuity of both eyes at presentation was 1/60. Grade 1 RAPD was present in right eye and pupil was normally reacting to light in left eye. Intraocular pressure was normal and Ocular motility was full in all gazes. Color Vision was poor andContrast Sensitivity: log 0.3 in both eyes. Visually Evoked Potential shows bilateral prolonged peak p100 latency (OD :145msOS :180.9ms).

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On slit lamp examination, there was no abnormality seen in lids/, conjunctiva and cornea. There was immature senile cataract NS2 in both eyes. Gonioscopy examination shows open angles in both the eyes.

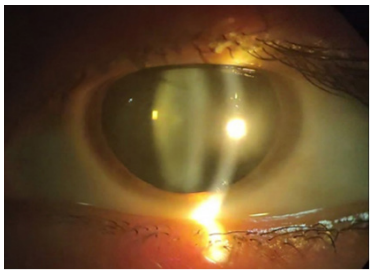


Figure a: Slit lampphotograph of right eye.

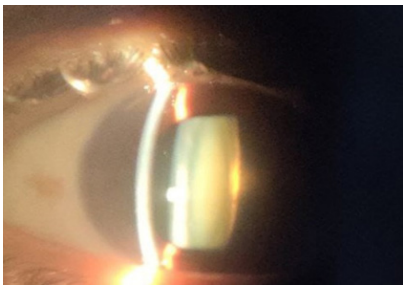


Figure b: Slit lampphotograph of left eye.

Fundus examination of the both the eyes revealsharply demarcated areas of chorioretinal atrophy in the periphery of temporal retinae. Some circular lesions that have coalesced to form large confluent areas. Numerous small yellow white punctate lesions are seen all over the background. Maculae are atrophied with hard exudates.

Optic disc of the right side shows C:D ratio of 0.6 with peripapillary atrophy around margins and subtle bayonetting sign.

Optic disc of the left eye shows C:D ratio of 0.9 with cupping with loss of ISNT rule, nasal shifting of blood vessels, bayonetting sign and barring of circumlinear vessels.

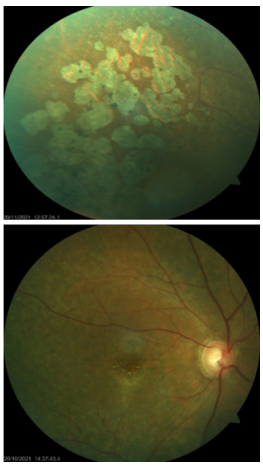


Figure c: Fundus picture of right eye.

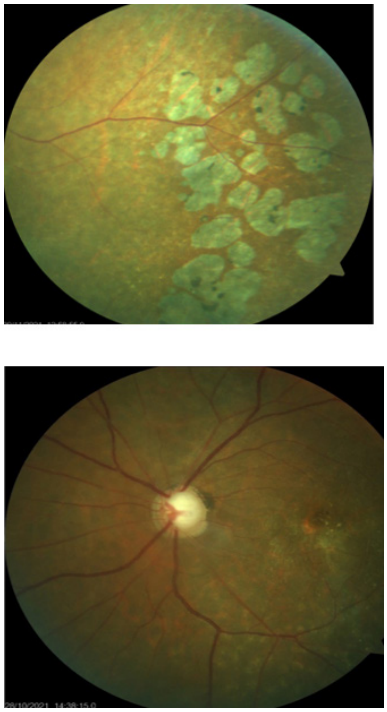
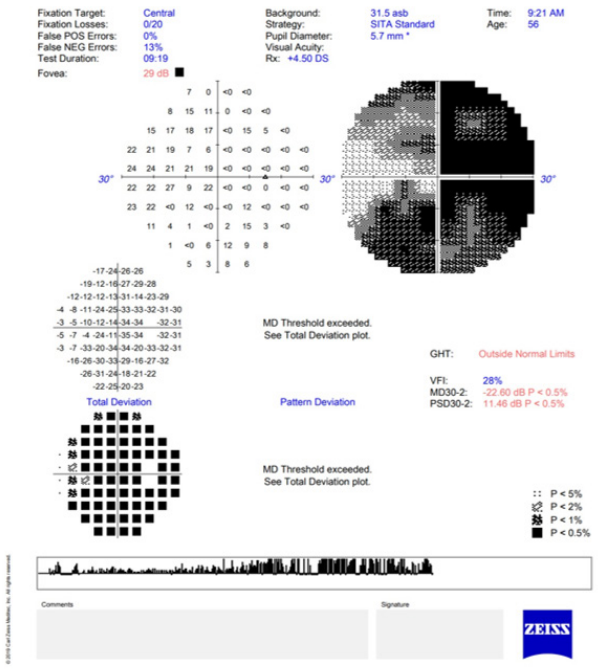


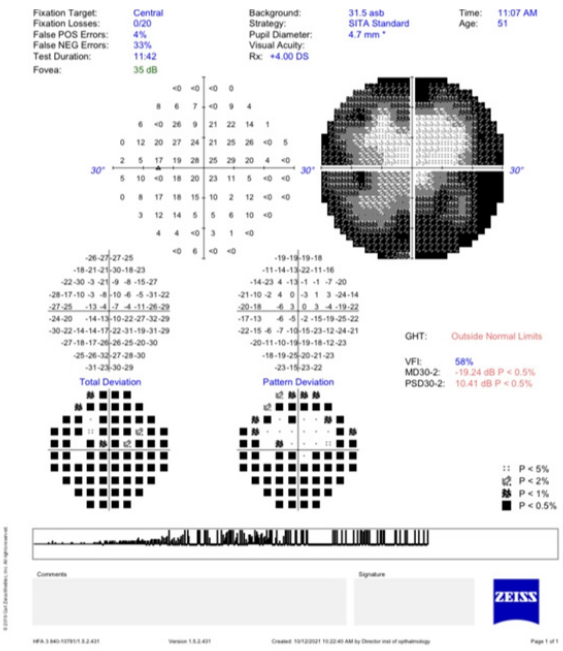
Figure d: Fundus Picture of Left Eye.

VISUAL FIELD ANALYSIS

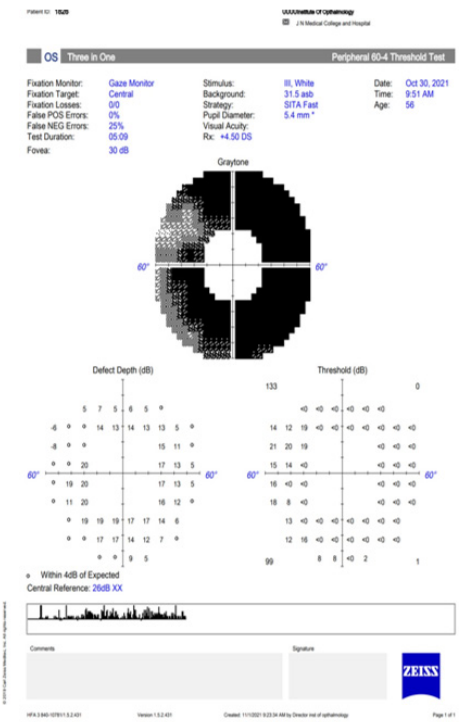
The 30-2 showing decreased foveal sensitivity in right eye. There is diffuse loss temporal and central visual fields in right eye and diffuse loss of inferior visual field in left eye.



Perimetry 30-2 of Right Eye



Perimetry 30-2 Left Eye.



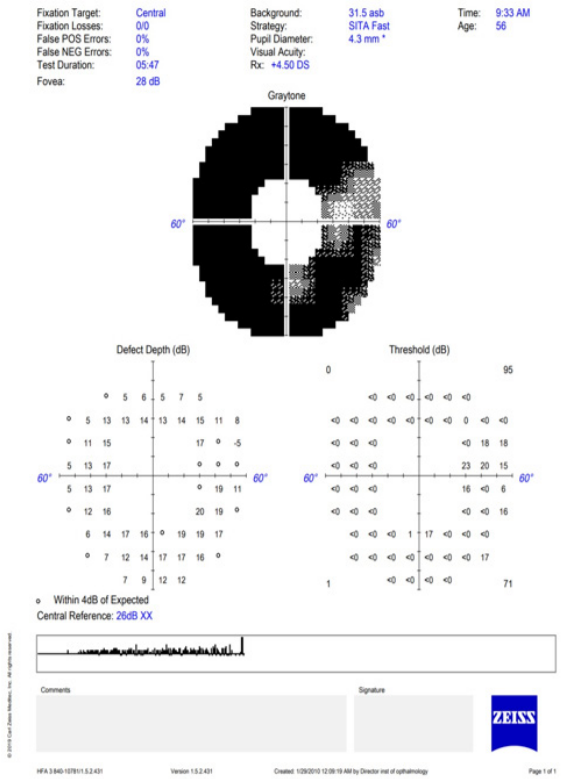
Peripheral 60-4 Left Eye.

OCT of right eye shows distorted foveal contour, presence of subretinal fluid with subretinal deposits which indicate towards old Central Serous Retinopathy.

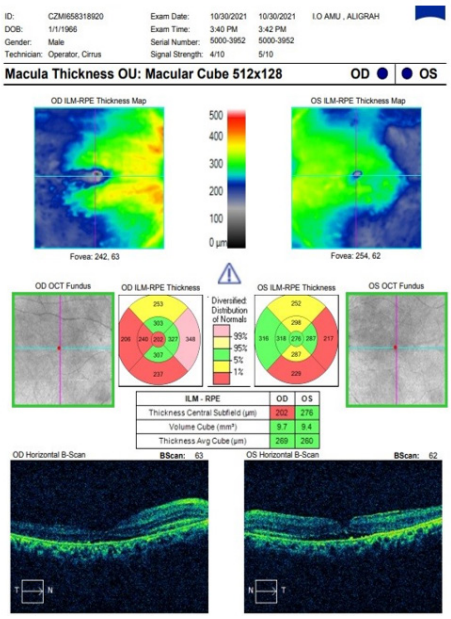
OCT of left eye shows presence of subretinal fluid with Central Serous Retinopathy with grade 3 macular hole.

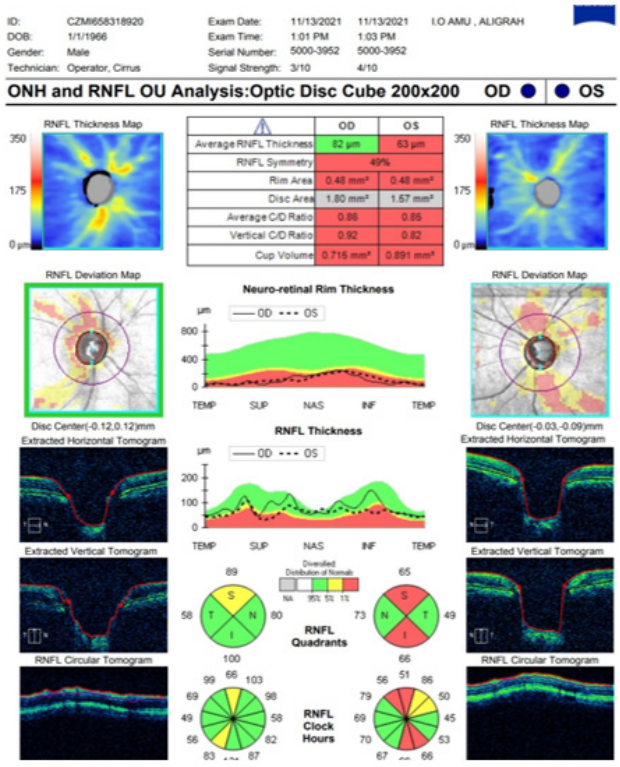
Borderline superior and inferior thinning of RNFL in left eye is seen.

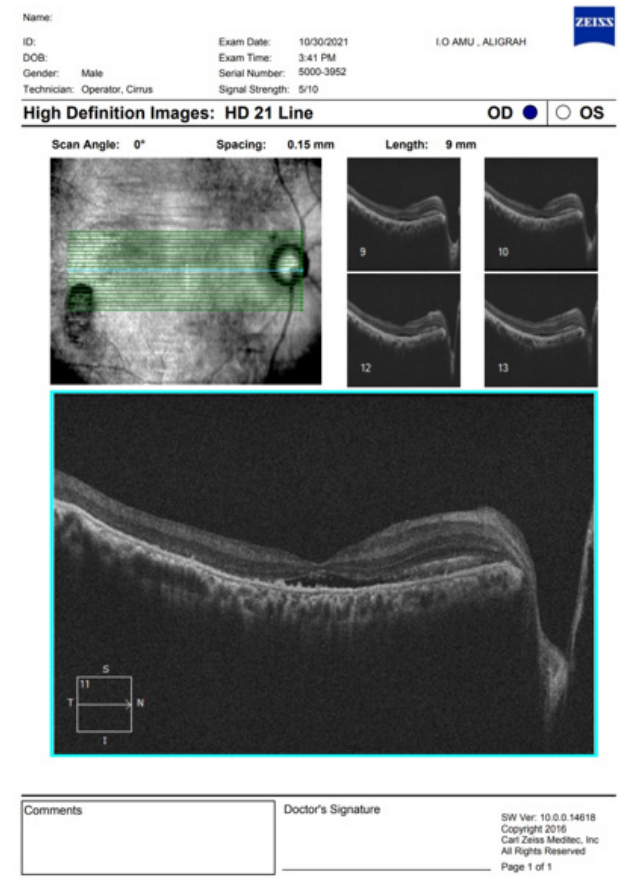
Bilateral thinning of ganglion cell layer is also seen.

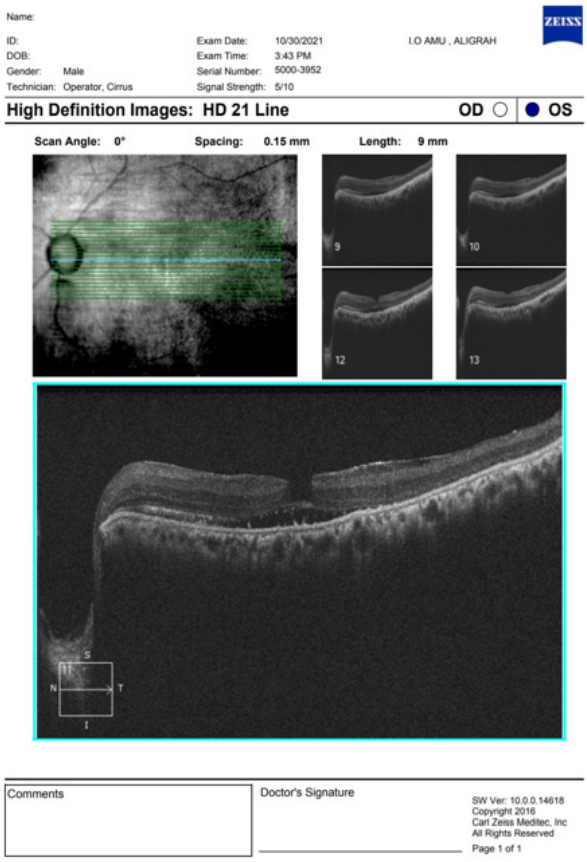


Peripheral 60-4 Right Eye.









DISCUSSION

Our patient presented with progressive diminution of vision in night and constriction of visual field due to progressive chorioretinal degeneration, progressive macular and glaucomatous changes and cataract formation that correlates with the progression of Gyrate atrophy.

Macular involvement occurs late in the disease Gyrate atrophy and macular complications like Cystoid macular edema, macular hole, foveal thinning, intraretinal cystic spaces, Vitreous haemorrhage and Retinal detachment are well documented with both Gyrate Atrophy and Fundus Albipunctatus.

OCT gives evidence of bilateral foveal thinning and bilateral CSR related maculopathy along with macular hole in left eye. All these are known complications of Gyrate Atrophy which lead to earlier and more severe loss of vision.

The optic disc and visual field changes along with gonioscopy examination indicate towards open angle glaucoma associated with gyrate atrophy and *Fundus Albipunctatus.*

CONCLUSION

Both Gyrate Atrophy and *Fundus Albipunctatus* are rare entities with the prevalence of the latter being unknown. Both

these entities occurring together along with glaucoma makes this case rarest of rare. The macular findings in this patient are already known to occur as their associates. Tab pyridoxine and arginine free diet should be advised to the patient along with management of glaucoma. Surgical management for cataract can also be considered. Family counselling becomes very important because of the known family history and inheritance pattern.

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Declaration of patient consent:

The author certify that they have obtained all appropriate patient consent. The patient has given consent for his images and other clinical information to be reported in the journal. They understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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