

Multimodal Imaging of Gyrate Atrophy Associated With Foveoschisis, Epiretinal Membrane and Optic Nerve Head Drusen: A Case Report

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ABSTRACT

We report a young male patient with gyrate atrophy (GA) complicated by foveoschisis, epiretinal membrane and optic nerve head drusen (ONHD) using multimodal imaging. A 25-year-old man presented with a history of progressive night blindness and visual impairment over six years. He had undergone cataract operation for both eyes in another hospital three months previously. He had a history of high myopia. His best corrected visual acuity (BCVA) was 6/20 on Snellen chart in both eyes. Anterior segment examination showed presence of bilateral intraocular lens. Dilated fundus examination revealed bilateral sharply-defined and confluent chorioretinal atrophic areas in the mid-peripheral zone. Large optic disc and temporal crescent were compatible with high myopia. The patient underwent imaging with colour fundus photography, fundus autofluorescence (FAF), fundus fluorescein angiography (FFA), optical coherence tomography (OCT). After all these evaluations, GA associated with foveoschisis, epiretinal membrane and ONHD were detected. Elevated levels of plasma ornithine were also detected (520 micromol/L) to confirm diagnosis. Because of possible complications, biomicroscopic examination with the multimodal imaging must be done carefully to detect the problems associated with gyrate atrophy.

Key Words: Gyrate atrophy, foveoschisis, epiretinal membrane, optic nerve head drusen.

INTRODUCTION

Gyrate atrophy (GA) is a rare, autosomal recessive metabolic disorder characterized by sharply demarcated and gradually coalesced circular or oval areas of chorioretinal atrophy. First of all, atrophy is observed in the-mid periphery of fundus and then spreads anteriorly and posteriorly.^{1,2} In GA caused by the deficiency of mitochondrial enzyme ornithine-delta-aminotransferase (OAT), which results from hyperornithinemia, is elevated up to 20-folds above normal levels.³ Its treatment involves restricting the intake of arginine, which is the precursor of ornithine, and using pyridoxine supplements to increase the OAT activity.⁴ The disease begins with deficiency in peripheral and night vision during the first decade of life. Macular and central visions are affected in the 4th to 5th decades.⁵

GA is associated with high myopia, subcapsular cataract, optic atrophy, macular edema, vasculitis, and rarely

retinal detachment, optic nerve head drusen (ONHD) and choroidal neovascularization.⁶⁻⁹

In this paper, we present a young male patient with GA complicated by foveoschisis, epiretinal membrane, and ONHD using colour fundus photography, fundus autofluorescence (FAF), fundus fluorescein angiography (FFA), optical coherence tomography (OCT).

CASE PRESENTATION

A 25-year-old man presented to our hospital with a history of progressive night blindness and visual impairment over six years. 3 months before, he had undergone cataract operation and intraocular lens implantation for both eyes in another hospital. He had a history of high myopia. Elevated levels of plasma ornithine were also detected (520 micromol/L) to confirm diagnosis.

Best corrected visual acuity (BCVA) was 6/20 on Snellen

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chart in both eyes with -1.0 D Sph, -1.0 D Cyl x 20° in the right eye and -1.0 D Sph, -1.25 Cyl x 140° in the left eye. Slit lamp biomicroscopic examination of anterior segment showed presence of bilateral intraocular lens and vitreous degeneration. Ophthalmoscopic examination of fundus showed bilateral sharply defined and confluent chorioretinal atrophic areas in the mid-peripheral zone. Large optic disc and temporal crescent were compatible with high myopia (Fig. 1a, b). Axial lengths measured by optical biometry (Lenstar LS 900, Haag-Streit AG, Köniz, Switzerland) were 26.52/26.27 mm (R/L eyes). Intraocular pressure was normal in both eyes.

OCT (RTVue XR100-2, Optuvue, Fremont, California, USA) showed hyporeflective spaces separated by multiple

vertical bridges, which suggested foveoschisis in the right eye (Fig. 2a) and epiretinal membrane in the left eye (Fig. 2b). The schisis cavities coalesced at the foveal center, forming rounded cyst like cavities. FFA (CF-1, Canon, Japan) illustrated confluent hyperreflective areas with prominent choroidal vasculature at the early and late phases in both eyes. Any leak at the macula did not appear even at the late phase in the right eye (Fig. 3a). The hyperfluorescence area in the inferior part of the macula was compatible with epiretinal membrane in the left eye (Fig. 3b). FAF (CF-1, Canon, Japan) showed drusen at the optic nerve head, while one was in the right eye (Fig. 4a) and two in the left eye (Fig. 4b).

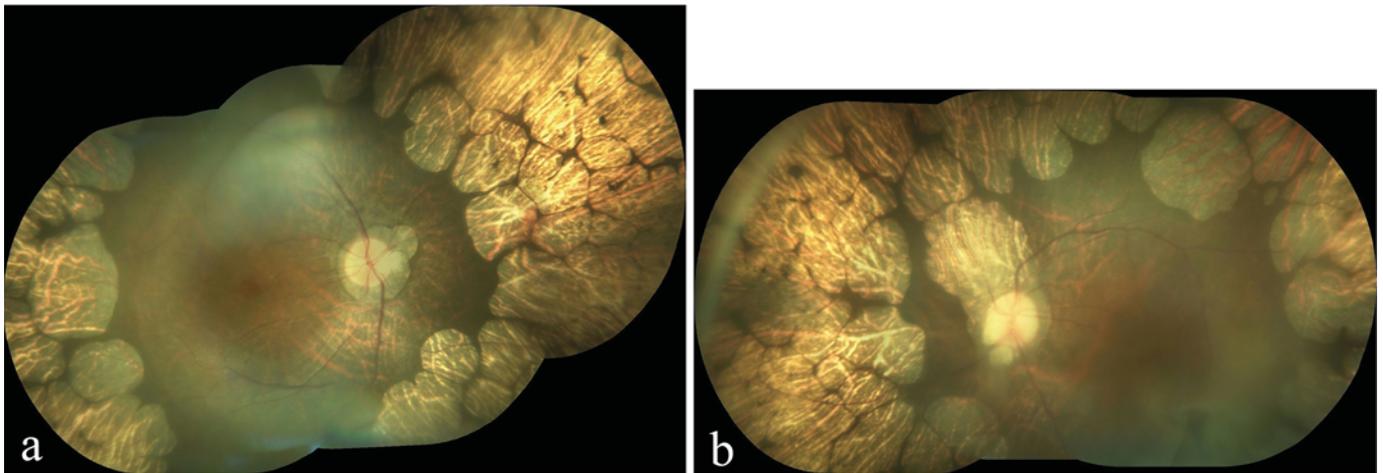


Figure 1. Fundus photographs of a gyrate atrophy patient, 25 years old. (a) Right eye. (b) Left eye.

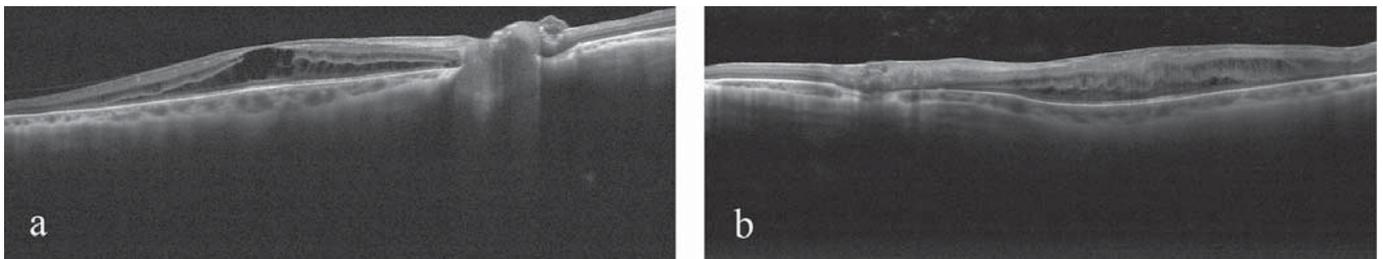


Figure 2. Optical coherence tomography images. (a) Foveoschisis at the right eye. (b) Epiretinal membrane at the left eye.

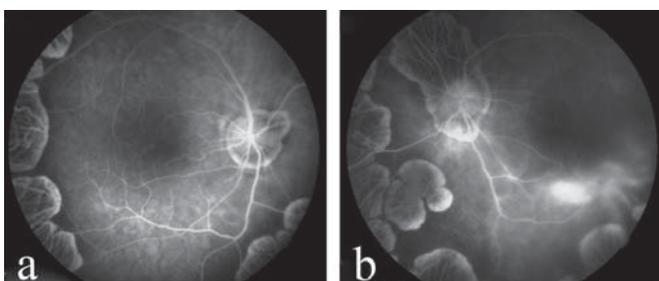


Figure 3. Fundus fluorescein angiography images (late phases). (a) Right eye. (b) Left eye.

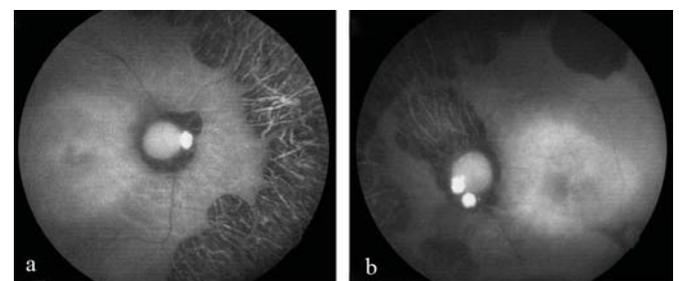


Figure 4. Fundus autofluorescence images. (a) Right eye. (b) Left eye.

DISCUSSION

The typical symptoms of GA include night vision problem and loss of peripheral visual field usually during first decade of life. Macula remains preserved till midlife. Diffuse chorioretinal atrophy, myopic changes and lens opacities are the most common causes of the visual problem in patients with GA.¹⁰ Macular changes including cystoid macular edema, epiretinal membrane and choroidal neovascular membrane have been reported to be present in this disease.¹¹⁻¹³ Two cases with a macular hole were published before by Tripathy et al. and Sharma et al.^{14,15} We observed an epiretinal membrane on the left eye of our patient.

High myopia is the most common cause of foveoschisis. Its pathogenesis is believed to be multifactorial including anterior traction by the vitreous and epiretinal membrane or retinal vascular traction and by a progressive ectasia of the sclera.^{16,17} Familial retinoschisis, Goldmann-Favre syndrome and enhanced S-cone syndrome are generalized retinal diseases in foveoschisis.¹⁸⁻²⁰ Tripathy et al. reported two children from the same family with three siblings with hyperornithinemia, gyrate atrophy and foveoschisis with OCT and without leak at the fovea in FFA.¹⁴ Our case report is the second to present macular foveoschisis associated with gyrate atrophy.

A study conducted by Vannas-Sulonen showed ONHD in 3 of 21 patients with GA, with two being bilateral and one unilateral.²¹ A 14-year-old boy was also reported with cystoid macular edema and unilateral disc drusen.⁶ Our patient had bilateral ONHD confirmed by FAF.

By increasing the clinical usage of OCT, macular pathologies can be defined well. For the diagnosis of foveoschisis, OCT scanning is much more reliable than clinical evaluation based on symptoms, biomicroscopy and FFA. The OCT appearances were first described in details by Takano and Kishi in 1999 on patients with high myopia.²² Foveoschisis is a slowly progressive condition leading to severe central visual loss in untreated cases.

ONHD are cellular deposits of calcium, amino and nucleic acids and mucopolysaccharides.²³ Although ONHD are normally asymptomatic, they are frequently associated with visual field defects as a result of mechanical stress.²⁴ Congestion of the nerve head may lead to impaired blood flow and predisposition to acute vascular events such as retinal vein or artery occlusion and anterior ischemic optic neuropathy.²⁵ In addition, chronic ischemia can result in subretinal neovascularization.²⁶

Our case report is the first to present the association between ONHD, foveoschisis and epiretinal membrane in GA patients. In conclusion, biomicroscopic examination

with the support of FFA, OCT and FAF must be performed carefully to detect the problems associated with GA due to possible complications.

Availability of data and material

All data generated or analyzed during this study is included in this published article.

Consent for publication

The patient gave a signed consent for the publication of his photography.

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REFERENCES

1. Micheal P, Eliot B. Diagnosis and Treatment of Gyrate Atrophy. *Int Ophthalmol Clin.* 1993; 33:229-36.
2. Takki KK, Milton RC. The natural history of gyrate atrophy of the choroid and retina. *Ophthalmology.* 1981;88:292-301.
3. Kaiser-Kupfer MI, Valle D, Del Valle LA. A specific enzyme defect in gyrate atrophy. *Am J Ophthalmol.* 1978;85:200-4.
4. Santinelli R, Costagliola C, Tolone C, et al. Low-protein diet and progression of retinal degeneration in gyrate atrophy of the choroid and retina: A twenty-six-year follow-up. *J Inher Metab Dis.* 2004;27:187-96.
5. Takki K. Gyrate atrophy of the choroid and retina associated with hyperornithinaemia. *Br J Ophthalmol.* 1974;58:3-23.
6. Goel N, Jain P, Arora S, et al. Gyrate Atrophy of the Choroid and Retina With Cystoid Macular Edema and Unilateral Optic Disc Drusen. *J Pediatr Ophthalmol Strabismus.* 2015;52:64.
7. Berbel RF, Rauen PI, Pallone RF, et al. Retinal detachment and gyrate atrophy of the choroid and retina: case report. *Arq Bras Oftalmol.* 2012;75:59-60.
8. Büyüktortop N, Alp MN, Sivri S, et al. Gyrate atrophy of the choroid and retina: a case report. *Turk J Pediatr.* 2011;53:94-6.
9. Surekha B, Bhandari A, Dhayதாக P, et al. Gyrate atrophy of choroid and retina with myopia, cataract and systemic proximal myopathy: A rare case report from rural India. *Australas Med J.* 2012;5:639-42.
10. Marano F, Deutman AF, Leys A, et al. Hereditary retinal dystrophies and choroidal neovascularization. *Graefes Arch*

- Clin Exp Ophthalmol. 2000;238:760-4.
11. Piozzi E, Alessi S, Santambrogio S, et al. Carbonic anhydrase inhibitor with topical NSAID therapy to manage cystoid macular edema in a case of gyrate atrophy. *Eur J Ophthalmol.* 2017; 8:179-83.
 12. Feldman RB, Mayo SS, Robertson DM, et al. Epiretinal membranes and cystoid macular edema in gyrate atrophy of the choroid and retina. *Retina.* 1989;9:139-42.
 13. Inanc M, Tekin K, Teke MY. Bilateral choroidal neovascularization associated with gyrate atrophy managed with intravitreal bevacizumab. *Int Ophthalmol.* 2018;38:1351-5.
 14. Tripathy K, Sharma YR, Chawla R, et al. Ultra-wide Field Imaging of an Operated Macular Hole in Gyrate Atrophy. *J Ophthalmic Vis Res.* 2016;11:336-7.
 15. Sharma YR, Singh DV, Azad RV, et al. Gyrate atrophy with bilateral full thickness macular hole. *Eye (Lond).* 2006;20:745-7.
 16. Gaucher D, Haouchine B, Tadayoni R, et al. Long-term Follow-up of High Myopic Foveoschisis: Natural Course and Surgical Outcome. *Am J Ophthalmol.* 2007;143:455-62.
 17. Ikuno Y, Gomi F, Tano Y. Potent retinal arteriolar traction as a possible cause of myopic foveoschisis. *Am J Ophthalmol.* 2005;139:462-7.
 18. Ozdemir H, Karacorlu S, Karacorlu M. Optical coherence tomography findings in familial foveal retinoschisis. *Am J Ophthalmol.* 2004;137:179-81.
 19. Fishman GA, Jampol LM, Goldberg MF. Diagnostic features of the Favre-Goldmann syndrome. *Br J Ophthalmol.* 1976;60:345-53.
 20. Marmor MF, Jacobson SG, Foerster MH, et al. Diagnostic clinical findings of a new syndrome with night blindness, maculopathy, and enhanced S cone sensitivity. *Am J Ophthalmol.* 1990;110:124-34.
 21. Vannas-Sulonen K. Progression of gyrate atrophy of the choroid and retina. A long-term follow-up by fluorescein angiography. *Acta Ophthalmol.* 1987;65:101-9.
 22. Takano M, Kishi S. Foveal retinoschisis and retinal detachment in severely myopic eyes with posterior staphyloma. *Am J Ophthalmol.* 1999;128:472-6.
 23. Lam BL, Morais CG, Pasol J. Drusen of the optic disc. *Curr. Neurol. Neurosci. Rep.* 2008;8: 404-8.
 24. Sato T, Mrejen S, Spaide RF. Multimodal imaging of optic disc drusen. *Am J Ophthalmol.* 2013;156:275-82.
 25. Silverman AL, Tatham AJ, Medeiros FA, et al. Assessment of optic nerve head drusen using enhanced depth imaging and swept source optical coherence tomography. *J Neuroophthalmol* 2014; 34: 198-205.
 26. Alkin Z, Ozkaya A, Yilmaz I, et al. A single injection of intravitreal ranibizumab in the treatment of choroidal neovascularisation secondary to optic nerve head drusen in a child. *BMJ Case Rep.* 2014;2:2014.