

Three siblings with gyrate atrophy of the choroid and retina: a case report

Maamouri Rym^{1,4}, Ferchichi Molka^{1,4}, Ben Chehida Amel^{2,4}, Hadj-Taieb Sameh^{3,4}, Cheour Monia^{1,4}

¹Department of Ophthalmology, Habib Thameur Hospital, Tunis 1069, Tunisia

²Department of Pediatrics, Rabta Hospital, Tunis 1007, Tunisia

³Laboratory of Biochemistry, Rabta Hospital, Tunis 1007, Tunisia

⁴University of Tunis El Manar, Faculty of Medicine of Tunis 1068, Tunisia

Correspondence to: Maamouri Rym. Department of Ophthalmology, Habib Thameur Hospital, Tunis 1069, Tunisia. ryma.maamouri@fmt.utm.tn

Received: 2023-07-20 Accepted: 2023-08-24

DOI:10.18240/ijo.2023.12.28

Citation: Rym M, Molka F, Amel BC, Sameh HT, Monia C. Three siblings with gyrate atrophy of the choroid and retina: a case report. *Int J Ophthalmol* 2023;16(12):2133-2135

Dear Editor,

We report the cases of three siblings with gyrate atrophy (GA) of the choroid and retina with foveoschisis, anterior subcapsular cataracts, and capsular bag contraction. GA is a rare autosomal recessive degenerative disorder of the choroid and retina. About one-third of all reported cases are from Finland where the incidence is estimated to be around 1:50 000 whereas the theoretical global incidence is only 1:1 500 000^[1]. GA results from mutations in the gene encoding for the enzyme ornithine-delta-aminotransferase (OAT). A decrease in the activity of the enzyme leads to an increase in the plasma levels of the amino acid ornithine which is believed to have a toxic effect on the retinal pigment epithelial cells^[1]. Although the progressive development of chorioretinal atrophic patches is the main finding in GA, other ocular manifestations such as myopia, macular cystic changes, and posterior subcapsular cataracts may occur^[1]. The majority of patients with GA have a visual acuity of less than 20/200 between 40 and 55 years of age^[2].

We hereby report the case of three members of a Tunisian consanguineous family with GA. Our purpose was to emphasize the risk of spontaneous lens dislocation and postoperative capsular fibrosis associated with this disease.

All procedures adhered to the tenets of Declaration of Helsinki. Oral and written consent was obtained by the three patients.

Case 1 A 41-year-old female with no past medical history presented to our department with a chief complaint of bilateral progressively worsening blurring of vision. She was born to a first-degree consanguineous marriage and had 7 siblings. Her ophthalmic history revealed axile myopia and bilateral posterior subcapsular cataract. She had undergone 14mo earlier consecutive uneventful phacoemulsification surgeries with implantation of soft hydrophobic acrylic intraocular lenses (IOL) in the capsular bag. Upon presentation, her best-corrected visual acuity (BCVA) was 20/63 in both eyes. Intraocular pressure was within the normal range. Slit-lamp examination revealed the presence of in-the-bag IOLs in both eyes with capsular contraction in her right eye (OD; Figure 1A) and secondary cataract in her left eye (OS; Figure 1B). Dilated fundus examination showed bilateral extensive confluent patches of chorioretinal atrophy with sharply defined borders in the midperiphery of the retina sparing the macula. Areas of hyperpigmented clumps were adjacent to these patches (Figure 2A, 2B). Swept-source optical coherence tomography (SS-OCT) of the macula revealed the presence of bilateral foveoschisis (Figure 2C, 2D). Blood ornithine level was 361 $\mu\text{mol/L}$ (normal: 2-186 $\mu\text{mol/L}$), thus confirming the diagnosis of GA. The patient was started on an arginine-restricted diet and vitamin B6 supplementary therapy.

Case 2 A 47-year-old male, an older sibling of patient 1, presented to our department for a systematic examination following his sister's diagnosis. He had no known medical history and has never consulted an ophthalmologist. His BCVA was 20/200 in OD and counting fingers at 50 centimeters in OS. Slit-lamp examination showed bilateral posterior subcapsular cataracts with phacodonesis in OD (Figure 3A, 3B). No history of ocular trauma and no signs of pseudoexfoliation were found. Dilated fundus examination revealed confluent areas of well-demarcated chorioretinal atrophy sparing the macula. Axial length measured with interferometry was 28.39 mm and 28.05 mm in OD and OS, respectively. Macular SS-OCT revealed the presence of bilateral foveoschisis. Due to lack of blood and genetic testing, a presumptive diagnosis of GA was made by relying

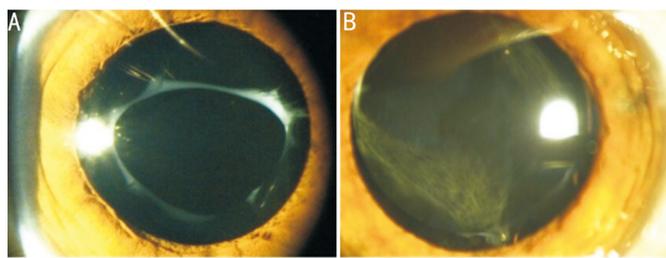


Figure 1 Anterior segment photographs of patient 1 showing contraction of the anterior capsule rim in her right eye (A) and opacification of the posterior capsule in her left eye (B).

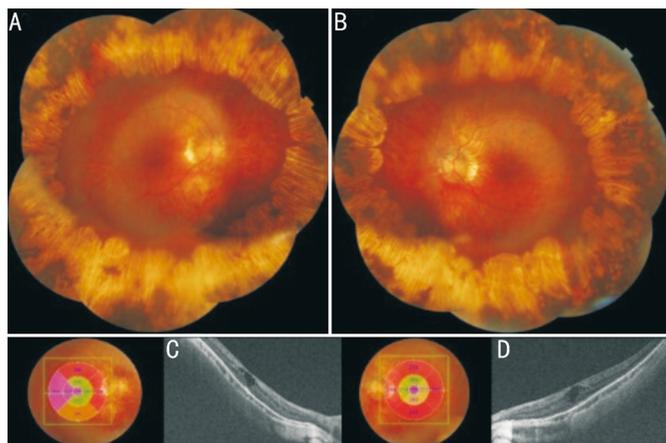


Figure 2 Multimodal imaging of patient 1 A, B: Fundus photographs showing in both OD (A) and OS (B) well-demarcated patches of chorioretinal atrophy in the mid-periphery sparing the macula, with visible choroidal vessels underneath and hyperpigmented clumps. Peripapillary atrophy, narrow retinal vasculature, and loss of the normal foveal reflex are also seen in both eyes. C, D: SS-OCT scans of OD (C) and OS (D) revealing central retinal thickening and hyporeflective spaces in the inner nuclear layer consistent with foveoschisis. OD: Right eye; OS: Left eye; SS-OCT: Swept-source optical coherence tomography.

on clinical signs and family history. He was started on an arginine-restricted diet and vitamin B6 supplementary therapy.

Case 3 A 46-year-old male, another sibling of patients 1 and 2, also presented to our department following his sister's diagnosis. His medical history was significant for a behavioral disorder. He had also undergone a previous phacoemulsification surgery with IOL implantation in OD. His BCVA was 20/200 in OD and counting fingers at 50 centimeters in OS. Slit-lamp examination of OD showed in-the-bag IOL with a wrinkled posterior capsule (Figure 3C). Examination of OS revealed an anterior subcapsular cataract with phacodonesis (Figure 3D). No history of ocular trauma and no signs of pseudoexfoliation were found. Dilated fundus examination showed patches of chorioretinal atrophy in the mid-periphery of the retina consistent with GA. Axial length measured with interferometry was 29.20 mm and 28.29 mm in OD and OS, respectively. Macular SS-OCT revealed the

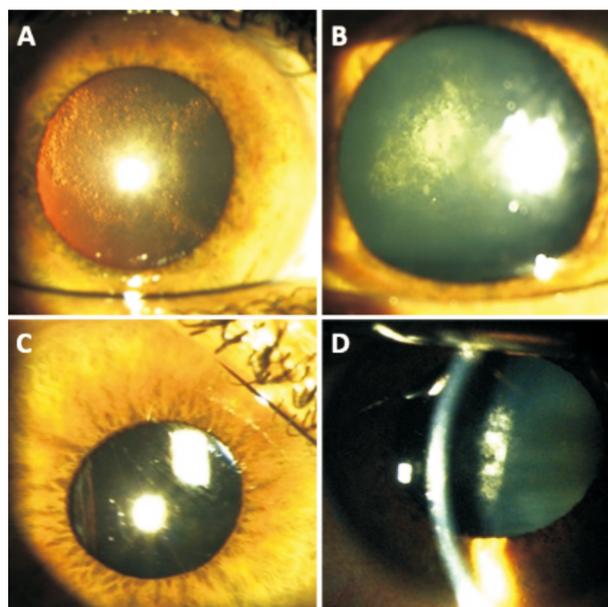


Figure 3 Anterior segment photograph A, B: Patient 2 showing posterior subcapsular cataracts in both OD (A) and OS (B). C, D: Patient 3 showing an in-the-bag intraocular lens with wrinkling of the posterior capsule in OD (C) and an anterior subcapsular cataract in OS (D). OD: Right eye; OS: Left eye.

presence of bilateral foveoschisis. Similarly, to case 2, a presumptive diagnosis of GA was made because of the lack of means to undergo blood or genetic testing. The patient was started on a diet with supplementary therapy and referred to his psychiatrist with the new updates regarding his diagnosis.

Cataracts in GA often develop during the second decade starting typically as punctuate lens opacities that develop along the posterior sutures and progress by spreading beneath the capsule^[3]. Capsular contraction after cataract surgery has been reported in a patient with GA who had bilateral in-the-bag dislocation of IOLs in the anterior chamber more than 15y after extracapsular cataract extraction. Significant fibrosis and opacification of the capsular bag were present. The authors suggested that capsular contraction may have led to the rupture of a portion of the zonular fibers causing IOL dislocation^[4]. Patient 1 in our series has developed contraction of the capsular bag rim and needs to be regularly examined in order to detect any subluxation of the IOL. Another case of spontaneous IOL dislocation more than 9y after phacoemulsification has been reported. Unlike the previous case, no signs of capsular fibrosis were found. The authors hypothesized the existence of zonular weakness similarly to what was described in some connective tissue disorders^[5]. Both patients 2 and 3 in our series had spontaneously subluxated lenses which may confirm this hypothesis. We suggest that they should be operated by experienced surgeons who must try to keep the strain on the zonules as minimal as possible. Thorough cortical clean-up, meticulous polishing under the anterior capsule. The use

of hydrophobic acrylic IOLs is also recommended to avoid capsular contraction and secondary cataracts. These patients should be regularly examined post-operatively in order to detect any complications and indicate the use of neodymium-doped yttrium aluminium garnet at an early stage.

Macular involvement in GA includes intraretinal cystic spaces, foveoschisis, cystoid macular edema, epiretinal membrane, macular hole, choroidal neovascularization and foveal thinning^[2,6]. Foveoschisis associated with GA is relatively rare and has already been described^[2,7]. Its pathogenesis is poorly understood. In addition to high axial length that induces retinal stretching, atrophy of the retinal pigment epithelium in areas of chorioretinal atrophy could influence the regulation of fluid balance leading to foveoschisis. However, Tekin^[8] described a GA-related foveoschisis with low myopia which may suggest the existence of other factors.

Apart from the characteristic ophthalmic findings in GA, several extra-ocular manifestations have been associated with this disorder mainly involving the central nervous system and the skeletal muscles^[1]. High levels of ornithine are believed to cause these symptoms^[2]. Through this case series, we wanted to highlight some unusual ophthalmic findings in patients with GA and to remind the ophthalmologist to look for extra-ocular manifestations associated with this disease.

ACKNOWLEDGEMENTS

Conflicts of Interest: Rym M, None; Molka F, None; Amel

BC, None; Sameh HT, None; Monia C, None.

REFERENCES

- 1 Montioli R, Bellezza I, Desbats MA, Borri Voltattorni C, Salviati L, Cellini B. Deficit of human ornithine aminotransferase in gyrate atrophy: molecular, cellular, and clinical aspects. *Biochim Biophys Acta Proteins Proteom* 2021;1869(1):140555.
- 2 Elnahry AG, Tripathy K. *Gyrate Atrophy of The Choroid and Retina*. StatPearls. Treasure Island (FL): StatPearls Publishing. 2023. <https://www.ncbi.nlm.nih.gov/books/NBK557759/>
- 3 Kaiser-Kupfer M, Kuwabara T, Uga S, Takki K, Valle D. Cataract in gyrate atrophy: clinical and morphologic studies. *Invest Ophthalmol Vis Sci* 1983;24(4):432-436.
- 4 Tsilou E, Rubin BI, Abraham FA, Kaiser-Kupfer M. Bilateral late posterior chamber intraocular lens dislocation with the capsular bag in a patient with gyrate atrophy. *J Cataract Refract Surg* 2004;30(7):1593-1594.
- 5 Kinori M, Desatnik H, Moisseiev J. Bilateral spontaneous dislocation of posterior chamber intraocular lens in a patient with gyrate atrophy. *Indian J Ophthalmol* 2012;60(2):133-134.
- 6 Gaudric A, Audo I, Vignal C, Couturier A, Boulanger-Scemama É, Tadayoni R, Cohen SY. Non-vasogenic cystoid maculopathies. *Prog Retin Eye Res* 2022;91:101092.
- 7 Guan W, Wang G. Partial regression of foveoschisis following vitamin B6 supplementary therapy for gyrate atrophy in a Chinese girl. *BMC Ophthalmol* 2021;21(1):93.
- 8 Tekin K. A rare association of foveoschisis with gyrate atrophy. *Delhi J Ophthalmol* 2017;28(1).

CORRIGENDUM

Artificial intelligence-assisted pterygium diagnosis: current status and perspectives

Bang Chen, Xin-Wen Fang, Mao-Nian Wu, Shao-Jun Zhu, Bo Zheng, Bang-Quan Liu, Tao Wu, Xiang-Qian Hong, Jian-Tao Wang, Wei-Hua Yang

(*Int J Ophthalmol* 2023;16(9):1386-1394. DOI:10.18240/ijo.2023.09.04)

The authors would like to make the following change to the above article:

Co-first authors: Bang Chen and Xin-Wen Fang

The authors apologize for any inconvenience caused by this error.