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Gyrate Atrophy of the Choroid and Retina: A Case Report

El Houssaine Ait Lhaj[1*], Said Alilou[2], Farah Belkadri[1], Assyia Lamkhoudem[1], Iliyas Elourdani[1], Salaheddine Bouabbadi[1], Fouad El Asri[1]

[1]Department of Ophthalmology, Avicenne Military Hospital, Marrakech, Morocco
[2]Eye Light Clinic, Agadir, Morocco

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***Corresponding author:** El Houssaine Ait Lhaj
Department of Ophthalmology, Avicenne Military Hospital, Marrakech, Morocco

Abstract | **Case Report**

Gyrate atrophy (GA) of the choroid and retina is a rare, autosomal recessive metabolic disorder caused by a deficiency of the mitochondrial enzyme ornithine aminotransferase. This report describes the clinical characteristics, multimodal imaging findings, and therapeutic outcomes in a 28-year-old Moroccan male from a first-degree consanguineous union who presented with a lifelong history of progressive nyctalopia and high myopia. Best-corrected visual acuity was 20/60 in the right eye and 20/100 in the left. Funduscopy revealed pathognomonic "circumferential garland" chorioretinal atrophic lacunae with scalloped margins and peripapillary atrophy. Multimodal imaging via fundus autofluorescence confirmed profound hypo-autofluorescence in atrophic zones, while infrared retinography highlighted well-demarcated, hyper-reflective scalloped patches. Spectral-domain optical coherence tomography identified significant cystoid macular edema (CME) predominantly within the outer retina. Laboratory analysis confirmed a pathognomonic hyperornithinemia of 950 µmol/L. Following management with a low-arginine diet, vitamin B6, and oral acetazolamide, the six-month follow-up showed a reduction in plasma ornithine levels to 252 µmol/L, correlating with a one-line improvement in visual acuity and stabilization of the macular status. This case underscores the utility of multimodal imaging in diagnosing GA and identifying treatable macular complications like CME, which remain significant obstacles to visual rehabilitation.

Keywords: Gyrate atrophy, Autosomal recessive metabolic disorder, Consanguinity, Ornithine aminotransferase deficiency, Hyperornithinemia, Multimodal imaging, Scalloped chorioretinal atrophy, Cystoid macular edema, Low-arginine diet, Vitamin B6.

INTRODUCTION

Gyrate atrophy (GA) represents a rare clinical entity inherited in an autosomal recessive fashion, fundamentally driven by a deficit in the mitochondrial enzyme ornithine aminotransferase (OAT) [1]. Since its initial characterization by Fuchs in the late 19th century [2], this metabolic disorder has been recognized for its progressive destruction of the chorioretinal architecture [1, 3]. The molecular basis involves mutations in the OAT gene on chromosome 10q26, which precipitates systemic hyperornithinemia [1, 4, 5]. Although the OAT enzyme is ubiquitously expressed in most human tissues, the pathological consequences are especially concentrated in the eye, manifesting as characteristic scalloped chorioretinal lesions [1]. We present the clinical characteristics and multimodal imaging findings of gyrate atrophy in a young Moroccan adult.

CASE REPORT

We report the case of a 28-year-old male, the eldest child of a first-degree consanguineous union, presenting with progressive nyctalopia and visual decline since childhood, which accelerated during his third decade. The patient was a known high myope (RE: -6.25 D; LE: -7.25 D) with a Best-Corrected Visual Acuity (BCVA) of 20/60 and 20/100, respectively. While the anterior segment was unremarkable, funduscopy revealed a striking "circumferential garland" of confluent atrophic chorioretinal lacunae with scalloped margins and peripapillary atrophy (Figure 1). To further characterize these findings, a multimodal imaging protocol was initiated. Fundus autofluorescence (FAF) demonstrated profound hypo-autofluorescence within the atrophic zones, reflecting total retinal pigment epithelium (RPE) loss (Figure 2), while infrared retinography highlighted these areas as well-demarcated, hyper-reflective scalloped patches (Figure 3).

Furthermore, SD-OCT identified foveal thickening with cystoid macular edema (CME) predominantly within the outer retina (Figure 4). Systemic evaluation revealed mild cognitive impairment, delayed language development, and speech disorders. While neuroimaging was not performed, plasma amino acid analysis confirmed a pathognomonic hyperornithinemia of 950 µmol/L (normal: 40–100 µmol/L). Based on the pathognomonic "garland" appearance and biochemical confirmation, a diagnosis of Gyrate Atrophy was established. Examination of the patient's parents and three siblings revealed no ophthalmological abnormalities. The patient was managed with a low-arginine diet, vitamin B6 supplementation (200 mg/day), and oral acetazolamide (125 mg twice daily). At the six-month follow-up, the plasma ornithine level had decreased to 252 µmol/L, correlating with a one-line improvement in BCVA and stabilization of the macular status.

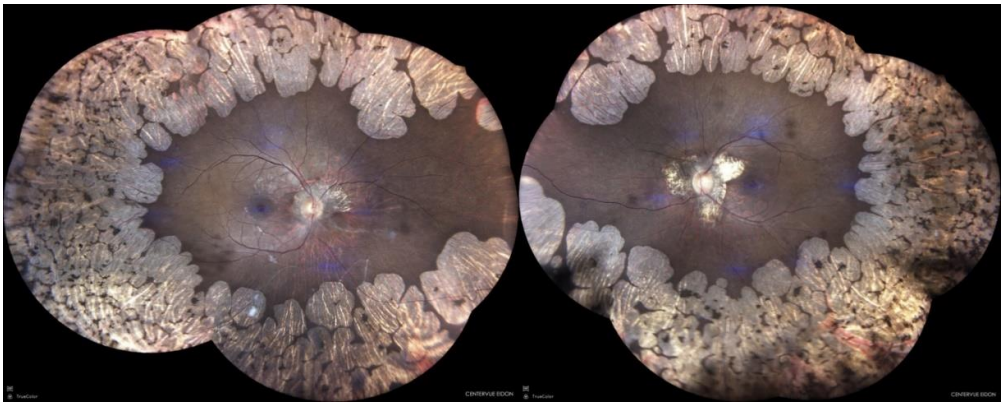

Figure 1: Color fundus photograph showing typical "circumferential garland" atrophic patches with scalloped margins in the mid-periphery and peripapillary area

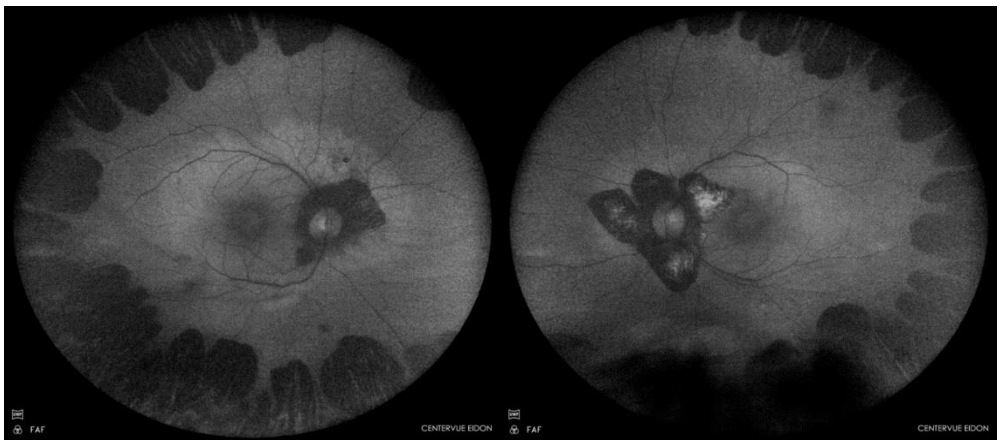

Figure 2: Fundus autofluorescence (FAF) image demonstrating areas of profound hypo-autofluorescence corresponding to total loss of the retinal pigment epithelium (RPE) and overlying photoreceptors

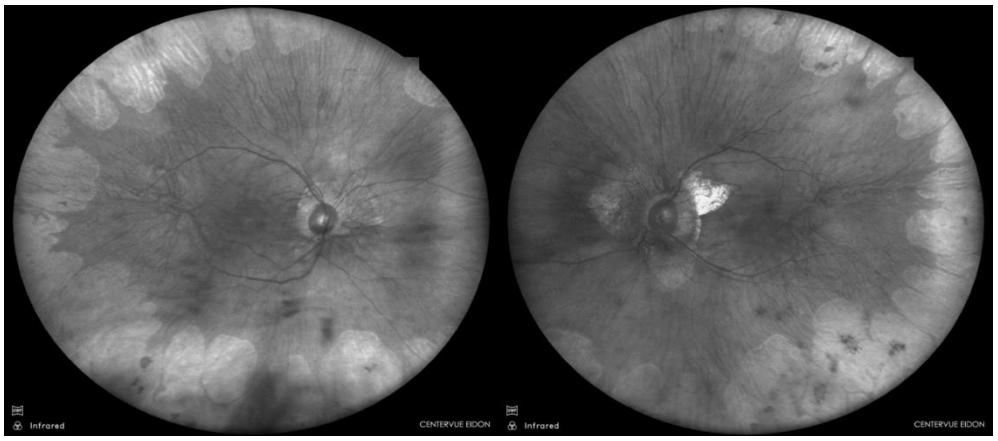

Figure 3: Infrared (IR) retinography highlighting well-demarcated, hyper-reflective scalloped lacunae, facilitating high-contrast visualization of the degenerative borders

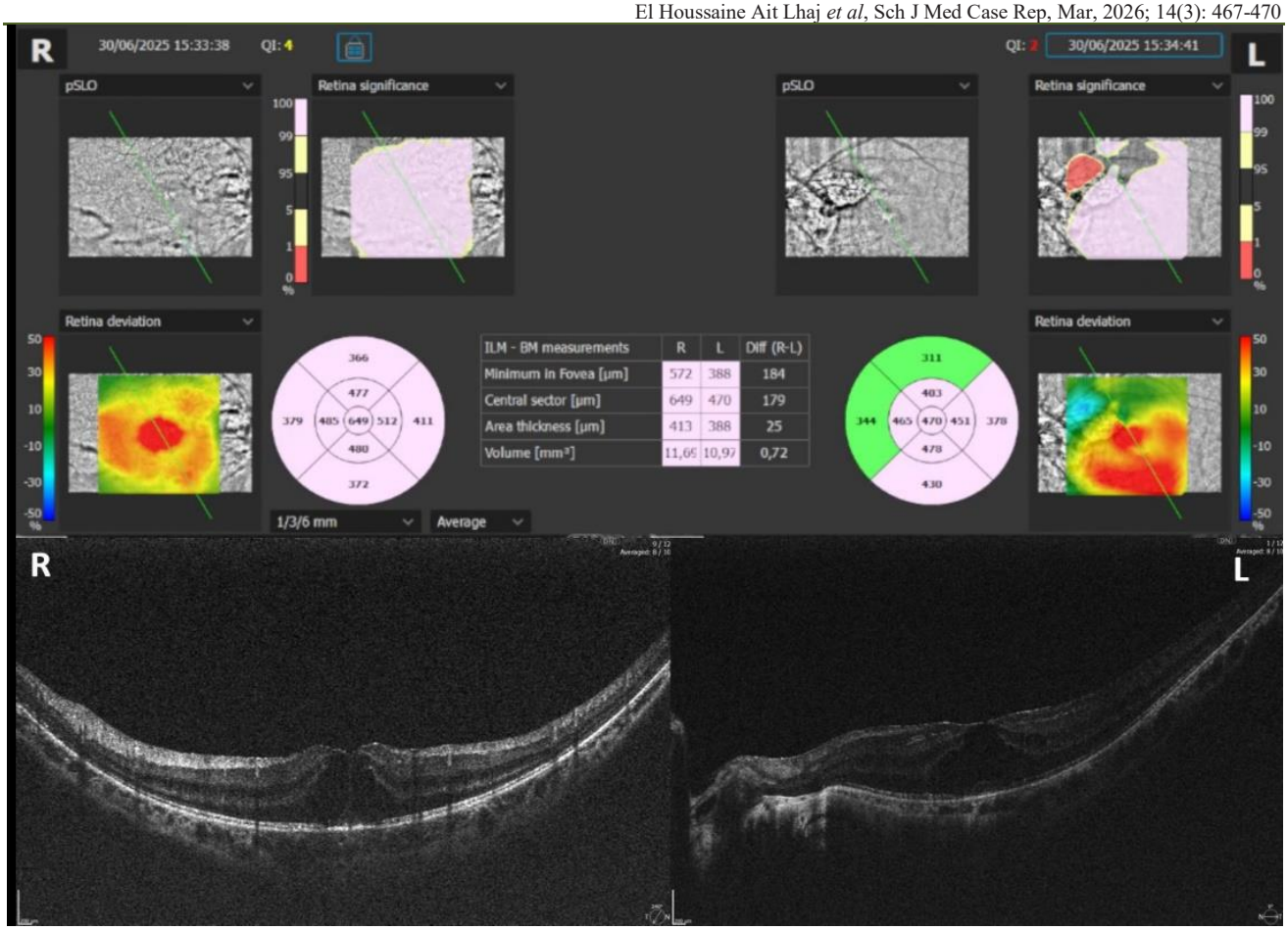

Figure 4: Spectral-Domain Optical Coherence Tomography (SD-OCT) cross-section through the fovea showing foveal thickening and cystoid macular edema (CME) predominantly involving the outer retinal layers

DISCUSSION

Gyrate atrophy (GA) of the choroid and retina is a rare, autosomal recessive metabolic disorder. It typically manifests with nyctalopia and progressive myopia, followed by a concentric reduction of the visual field secondary to peripheral chorioretinal loss [1, 6]. While GA is highly prevalent in Finland due to a founder effect, cases have been documented globally, including recent reports from North Africa (Tunisia, Egypt, and Morocco), where consanguinity may play a role in its incidence [1, 3, 6, 7].

GA is characterized by a profound systemic elevation of ornithine, typically exceeding normal physiological thresholds by ten to twenty times. This biochemical imbalance appears specifically cytotoxic to the retinal pigment epithelium (RPE) [1, 5]. Current evidence suggests that the RPE serves as the primary site of injury, with secondary atrophy of the choriocapillaris and photoreceptor complex following as the disease progresses [7, 8]. The structural hallmark of this process is the abrupt transition between healthy tissue and atrophic zones, creating the characteristic scalloped, "garland" borders.

While many authors report an early association with posterior subcapsular cataracts [1, 6], our patient presented with transparent lenses, underscoring that lenticular changes are common but not universal. However, a progressive decline in central visual acuity is frequently associated with macular complications of the disease, including intraretinal cystoid macular edema (CME), foveal detachment, epiretinal membrane, and geographic atrophy [1].

Beyond the eye, GA is a systemic metabolic disorder. The deficiency in the mitochondrial OAT enzyme leads to neurotoxicity—potentially due to secondary creatine deficiency—manifesting in some patients as intellectual disability, speech disorders, or epilepsy [9]. Our case reinforces this systemic link, as the patient exhibited mild cognitive impairment and delayed language development.

Therapeutic management focuses on reducing systemic ornithine levels to arrest the progression of chorioretinal atrophy. This is achieved through a strict low-arginine diet, avoiding protein-rich foods such as nuts, seeds, seafood, and dairy [1, 4]. In a subset of patients known as “pyridoxine responders,” high-dose vitamin B6 supplementation can partially restore OAT enzyme activity [1, 5]. While our patient achieved a significant biochemical response—with ornithine levels dropping from 950 µmol/L to 252 µmol/L—visual acuity

improvement was modest (one line). This was likely due to the presence of chronic CME, as identified on SD-OCT. This highlights the importance of multimodal imaging in monitoring macular status, as CME remains a major, yet treatable, obstacle to visual rehabilitation in these patients [7, 8].

CONCLUSION

This case underscores the critical role of multimodal imaging in the early diagnosis and longitudinal monitoring of gyrate atrophy. The pathognomonic "garland" atrophic pattern, while clinically suggestive, is objectively characterized through FAF and infrared retinography, while SD-OCT remains essential for identifying secondary macular complications like CME. In populations with high rates of consanguinity, a multidisciplinary approach combining aggressive metabolic control and pharmacological management of macular edema is crucial to stabilizing visual function and addressing the systemic neurological manifestations of the disease.

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Authors' Declaration

The work presented is original and has been conducted by the named authors. It does not contain any material that has been plagiarized or previously published in any form.

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