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Gyrate Atrophy of the Choroid and Retina in a Young Patient: A Case Report

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Abstract **Case Report**

Gyrate atrophy of the choroid and retina is a rare autosomal recessive disorder characterized by progressive metabolic degeneration of the retina and choroid. The condition results from a deficiency in ornithine delta-aminotransferase (OAT); a mitochondrial matrix enzyme that requires pyridoxal phosphate as a cofactor. We report the case of a young patient with this rare condition.

Keywords: retina, gyrate, atrophy, choroid.

INTRODUCTION

Gyrate atrophy (GA) of the choroid and retina is a rare autosomal recessive disorder marked by progressive metabolic degeneration of the retina and choroid. This condition stems from a deficiency in ornithine delta(δ)-aminotransferase (OAT), a mitochondrial matrix enzyme dependent on pyridoxal phosphate [1]. The hallmark biochemical feature of OAT deficiency is hyperornithinemia, with systemic ornithine levels 10 to 20 times higher than normal in plasma, urine, cerebrospinal fluid, and aqueous humor. The OAT gene has been cloned and mapped to chromosome 10. Clinically, GA presents with progressive midperipheral atrophy of the choroid and retina, often exposing the sclera and leading to severely reduced visual acuity. [2]

CASE REPORT

A 24-year-old male patient presented with complaints of gradual visual loss and progressive night vision deterioration occurring over the past several years. The best corrected visual acuity was 4/10 in both eyes. The refractive error was-2;00 D in the right eye and -1.00 D in the left eye.

On examination, his fundus exhibited bilateral severe chorioretinal atrophy involving the midperiphery. Fundus fluorescein angiography revealed leakage at the margin of chorioretinal atrophy. Dynamic Goldmann perimetry demonstrated visual field constriction in both eyes.

On plasma amino acid analysis, he had markedly elevated plasma levels of ornithine. The clinical diagnosis of the patient was consistent with OAT deficiency and GA of the choroid and retina.

The patient was treated with vitamin B6 (pyridoxine) 300 mg daily and an arginine-restricted diet.

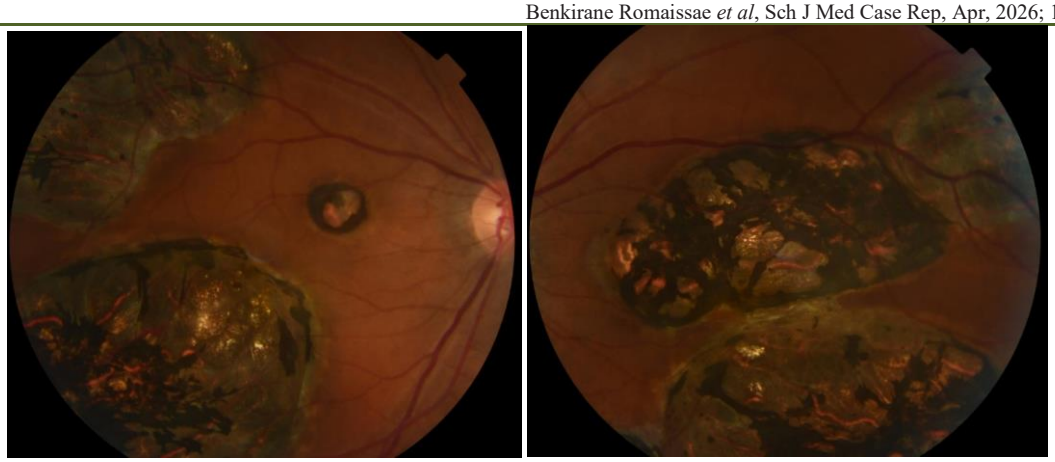

Figure 1: Retinography of the patient

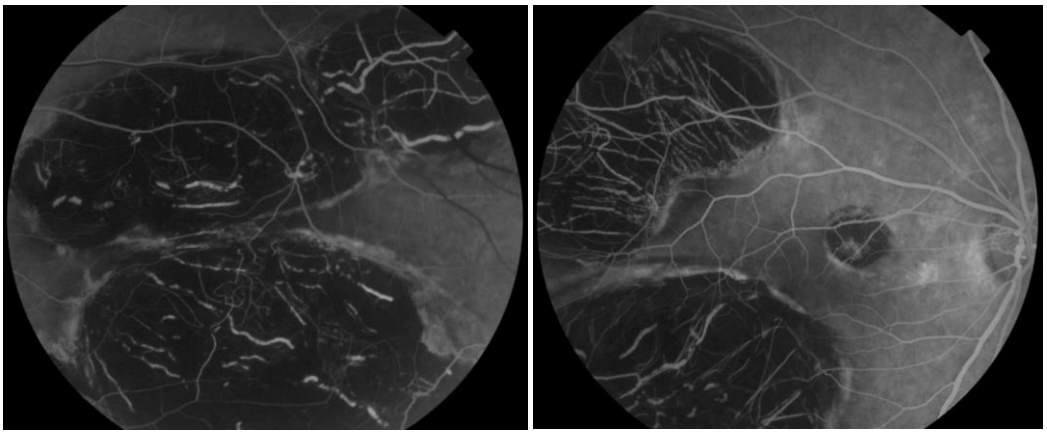

Figure 2: Angiography of the patient

DISCUSSION

Gyrate atrophy (GA) of the Choroid and Retina was first described by Fuchs in 1896 [3]. A rare autosomal recessive dystrophy characterized by progressive vision loss, particularly night blindness, along with myopia, subcapsular cataracts, and impaired dark adaptation. Patients typically develop central vision loss, leading to severe visual impairment or blindness by the fourth or fifth decade. Retinal examination reveals circular, well demarcated chorioretinal atrophy with hyperpigmented margins in the midperiphery, reduced electroretinogram (ERG) responses, and, in rare cases, vitreous hemorrhage [4,5].

While primarily an ophthalmic condition, GA has also been associated with mild intellectual disability, muscle weakness, straight hair, and neonatal failure to thrive [6-8]. However, our patient exhibited only visual symptoms without systemic involvement.

Diagnosis relies on markedly elevated plasma ornithine levels (5-20 times normal), often exceeding 400 µmol/L, accompanied by increased urinary excretion of lysine, arginine, and cysteine [9]. Genetic testing can confirm the diagnosis [10].

The metabolic basis of gyrate atrophy necessitates careful dietary management. Since arginine serves as the precursor for accumulating ornithine, we typically recommend restricting dietary arginine - primarily by limiting nuts, seeds, and other high-arginine foods. This approach has shown several important effects in clinical practice; First, the arginine-restricted diet reliably reduces plasma ornithine levels and appears to slow the progression of peripheral vision loss. Interestingly, the timing of intervention matters - patients who start the diet earlier in life tend to experience slower progression of their chorioretinal lesions [9,11]. However, it's crucial to note that this dietary modification doesn't reverse existing macular edema. We must balance these benefits against potential risks. Arginine remains an essential amino acid, particularly during growth and development. Overly restrictive diets can lead to complications like growth impairment and skin abnormalities. For this reason, we absolutely avoid arginine restriction in children under 3-4 years old [12].

In parallel with dietary management, we often trial pyridoxine (vitamin B6) supplementation. The response varies significantly between patients. While some studies report reductions in ornithine levels up to 50% with high doses (300-500 mg/day), our Finnish colleagues found that fewer than 5% of their patients responded meaningfully [13].

It should be stated that there are some limitations in this case report, such as we could not do the molecular analysis for our patient because of financial issues and the follow-up period was short for judging the long-term ophthalmologic results.

CONCLUSION

Gyrate atrophy (GA) of the fundus is a rare autosomal recessive disease characterized by deficiency of orni-thine-δ-aminotransferase (OAT). OAT deficiency causes hyperornithinemia, which results in progressive chorioretinal atrophy. [14]

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