

Case Report

## **An Unusual Presentation of Ornithine Aminotransferase Deficiency as Stroke with Gyrate Atrophy**

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### **ABSTRACT**

Gyrate atrophy (GA) of the choroid and retina is a rare autosomal recessive disease characterized by deficiency of ornithine- $\delta$ -aminotransferase (OAT). OAT deficiency results in progressive chorio retinal atrophy due to increase in plasma ornithine. Deficiency of a mitochondrial carrier protein that normally functions to transport ornithine into the mitochondria in the urea cycle results in Gyrate atrophy. We report a case of OAT deficiency in a 35 year old male who was admitted with stroke of one month duration and defective vision and progressive night blindness for fifteen years duration.

**Key words:** Gyrate atrophy, Ornithine aminotransferase deficiency, Hyperornithinemia, stroke, pyridoxine.

### **INTRODUCTION**

Gyrate atrophy (GA) of the choroid and retina was first described by Fuchs in 1896. [1] It is an autosomal recessive trait, with deficiency of pyridoxal phosphate-dependent mitochondrial matrix enzyme ornithine aminotransferase (OAT) which results in hyperornithinemia. [1]

Patients present with myopia, atrophy in the retina, loss of peripheral vision, and nyctolopia. [2] Though Gyrate atrophy usually does not affect intelligence, abnormalities may be observed in neurological examination and in brain imaging. It may also cause disturbances in peripheral nervous system, if type two muscle fibres are involved, mild weakness may result.

### **CASE REPORT**

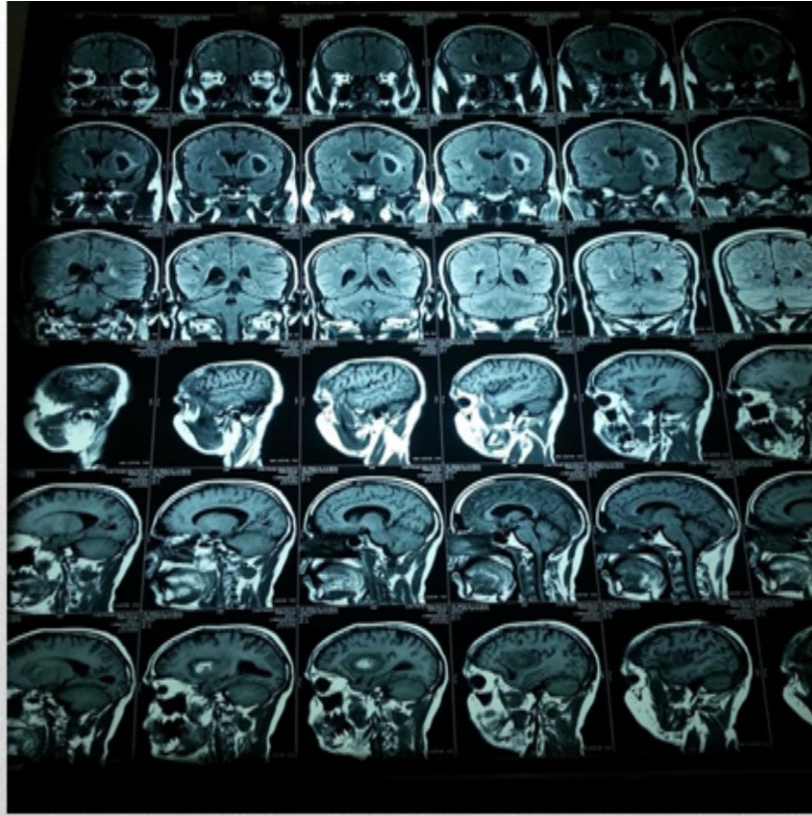
A 35 year-old male presented with history of one month old right facio

brachial monoparesis and gradual visual loss and progressive night vision deterioration of 15 years duration and without any significant medical comorbidities. His history was non contributory for any risk factors for stroke including hypertension. His brother also had history of gradual visual loss and night blindness.

On admission examination of Central Nervous System (CNS) showed right facio brachial monoplegia. Ophthalmic opinion was obtained which shows best corrected visual acuity (BCVA) with snellens chart as eyes 3/60., intraocular pressure right eye 19mmhg and left eye 17 mmhg. Colour visions of both eyes are defective. Both eye fundus shows media clear, disc pallor, vessels attenuation at arteries and veins, macula is normal and background retina shows bilateral hereditary pigmentary retinopathy with

peripheral and peripapillary chorioretinal atrophic areas with scalloping. (figure 2 and figure 3) The rest of examination was normal. His blood investigations including risk factor work up were normal. MRI showed an early sub acute haemorrhage in left lentiform nucleus with small vessel Ischaemic changes. Hence a clinical

diagnosis of Gyrate atrophy (GA) was considered. Although serum ornithine level measurements are required to establish the diagnosis, we performed a urine ornithine level in our patient due to cost constraints. The patient was found to have markedly elevated levels of urine ornithine, which confirmed the diagnosis.



**Figure 1:** MRI Brain showing early sub-acute hemorrhage in Lt lentiform nucleus with mild perilesional edema & Small vessel ischemic changes.



**Figure 2:** Fundus view of the right eye shows the peripheral and peripapillary chorioretinal atrophic areas with scalloping. Macula is spared.



**Figure 3:** Fundus view of the Left eye shows the peripheral and peripapillary chorioretinal atrophic areas with scalloping. Macula is spared.

## DISCUSSION

Gyrate atrophy (GA) of fundus is a rare degenerative disease. It is characterized by increase in ornithine levels in blood biochemically, due to deficiency of ornithine amino transferase. [3] McCulloch and Marliss have established that abnormal levels of ornithine is expressed in tissues including liver, retina, kidney, small intestine, muscle and splanchnic areas. [4] Gordon found a route of excess ornithine released in urine in Ornithine amino transferase (OAT) deficiency patients which suggests that methyl esterification of ornithine. [5]

OAT activity is reported to be high in the cell layer external to the photoreceptors in the retinal pigment epithelium. OAT deficiency is a progressive degenerative of choroid and retina with auto recessive pattern. By ophthalmoscopic appearance it is called as Gyrate atrophy of choroid and retina. [6] GA occurs due to phosphocreatine deficiency, hyperornithinemia, proline deficiency/ delta 1 pyrrolidine 5 carboxylate and excess polyamines. [5]

GA reports night blindness between ages 10-20 in paediatric age group which leads to blindness between ages 40-50. [7] Other ocular manifestations like posterior sub capsular cataracts, myopia and vitreous opacities. Neurologically patients may present with delayed language development and speech defects low intellectual activity, and mild proximal muscle weakness. Other features include alopecia, and bizarre elongated segmented mitochondria in liver biopsies. [8]

Improvement in visual acuity and electroretinograms in patients with gyrate atrophy has been reported after a low protein, arginine restricted diet. Administration of pyridoxine hydrochloride (Vitamin B6) leads to decreased activity of B6 dependent enzyme and stimulates OAT activity and results in reduction of plasma ornithine

levels. Treatment requires clinical and biochemical evaluation along with regular follow-ups. Enzyme replacement and gene therapy may prove to be promising treatment modalities for gyrate atrophy of choroid and retina in future. [9] To the best of our knowledge, this is the first case of Gyrate atrophy associated with haemorrhagic stroke reported.

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