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Original Research Paper

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GYRATE ATROPHY: A RARE CASE REPORT

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ABSTRACT

Background- Gyrate atrophy of the retina & choroid is a rare autosomal recessive inherited disease, characterized by progressive chorioretinal atrophy that results in progressive deterioration of peripheral & night vision leading to blindness.

Case presentation- This report presents a 40 year old woman consulting for a progressive fall of visual acuity RE with defective night vision. Ocular fundoscopy showed regions of confluent rounded chorioretinal atrophy. Visual field & retinal angiography were altered. High levels of plasma ornithine 685 nmol/mL) was detected and a diagnosis of gyrate atrophy of retina & choroid was made. The patient was treated with high dose pyridoxine supplement (300 mg/d for 6 months) & ornithine level of her serum was successfully reduced.

Conclusion- The exact mechanism of chorioretinal atrophy in hyperornithemia is not known and a small percentage of affected people respond to Vitamin B_6 supplimentation.

KEYWORDS :

INTRODUCTION:-

Gyrate atrophy (GA) of choroid & retina was first described by Fuchs in 1896. [1] Human hereditary deficiency of ornithine aminotransferse (OAT) activity is transmitted as an autosomal recessive trait, [2]and results in 10 to 20 fold increased level of plasma ornithine and is shown to be associated with GA. [3]The initial complaint of decreasing visual acuity & night vision is followed by the appearance of sharply demarcated, circular areas of chorioretinal atrophy with hyperpigmented margins in the mid- periphery of the fundus. This appears through the first 3 decades of life & leads to blindness in the 4th to 7th decades. Myopia, posterior subcapsular cataract, vitreous opacities may also be present. [4] Ornithine delta aminotransferase (OAT) is a mitrochondrial nuclear encoded pyridoxal phosphate enzyme that catalyzes the interconversion of ornithine glutamate & proline. Gyrate atrophy is a genetic disorder with increased frequency in Finnished population with an incidence of 1 case per 50,000 individuals in Finland.

We report a rare case of GA, in which the patient's high level of serum ornithine was responsive to therapy with Vitamin B6 dietary supplement & was reduced to near normal level.

Case report

A 40 year old woman presented with the complaint of gradual visual loss in right eye during the past 5 years. She had noticed night vision difficulties since the age of 20 years but did not seek medical advice. His visual acuity in RE was FC4M and in LE Was 6/6. Her best corrected visual acuity was 6/24 OD with a refractive error of -6.0 D sphere. Bilateral early posterior subcapsular cataract and vitreous degeneration. Fundus examination of right eye revealed peripapillary atrophy, severe arteriolar attenuation, sharply demarcated areas of choroid & retinal atrophy in gyrate shape with hyperpigmented patches involving the midperiphery and macula with dull foveal reflex, and left eye revealed peripapillary atrophy, severe arteriolar attenuation, sharply demarcated areas of choroid & retinal atrophy in gyrate shape with hyperpigmented patches involving the midperiphery, FR seen . Clinical diagnosis identified gyrate atrophy of choroid & retina. He had mild muscle weakness but his intelligence and electroencephalography (EEG) were normal. Aminoacid analysis revealed a high serum ornithine level (685 nmol /ml), with the normal range being 28-110 nmol/ml , Arginine, creatinine, lysine & glutamine levels in the serum were within the normal range. She was treated with Vitamin B6 at a dose of 300 mg/day for six months. This dietary supplement successfully reduced his serum ornithine level by more than 50% from 685 nmol/ml to 296 nmol/ml .

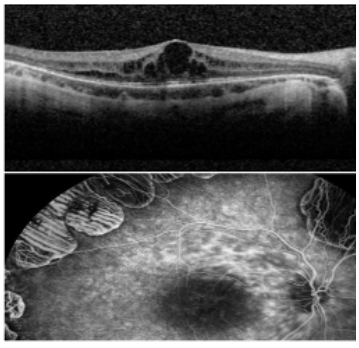

Figure 1: OCT shows macular oedema, and fundus angiography of gyrate atrophy.

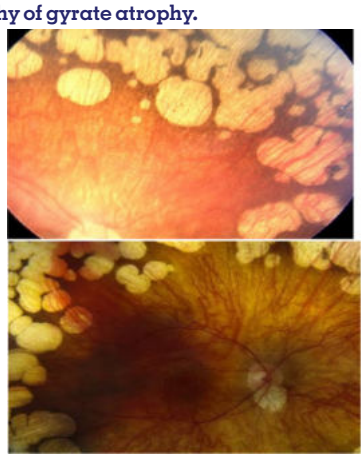

Figure 2: Fundus Photography - Fundus View Of The Eye Shows The Peripheral And Peripapillary Chorioretinal Atrophic Areas And Severe Arteriolar Attenuation.

DISCUSSION

OTA deficiency is characterized by progressive degeneration of the choroid & retina,[5] is inherited as an autosomal recessive pattern. In accordance with it's ophthalmoscopic appearance, it is called gyrate atrophy of choroid & retina and there is mild leakage of fluorescein at the margin of healthy appearing retina, where it abuts the gyrate lesions. Its main manifestation is a gradual loss of vision, night blindness & tunnel vision, with subsequent development of posterior

subcapsular cataract by 2nd decade,[6] progressive constriction of visual field & eventual loss of central vision in the 4th to 5th decade of life.[3] The mechanism of GA remains unknown; however, the adverse effects of creatinine or pyrroline-5-carboxilate (P5C) deficiency on retinal function are thought to be a causative factor.[7] The administration of pharmacologic doses of Vitamin B6 in a disorder caused by decreased activity of a B6- dependant enzyme is an established procedure.[7,8] Weleber and Kennaway in a clinical trial of Vitamin B6 for gyrate atrophy reported that 3 out of 7 patients responded to oral B6 supplementation with over 50% reduction of serum ornithine.[7,9] Our patient is a rare case of GA in that his serum ornithine level decreased by more than 50% from after administration of 300 mg/day Vitamin B6 as a dietary supplementation.

Conflict of interest - None

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