

Rare Presentation of Gyrate Atrophy in Pregnancy

A. M. Raja*, Siddharam Janti, C.Charanya Chendilnathan, Adnan Matheen

Department of ophthalmology, Chettinad Hospital and Research Institute, Chennai, India

*Corresponding Author:

Name: Dr. A. M. Raja

Email: dramraja@yahoo.com

Abstract: 34years pregnant female came with complaints of defective vision for two years and had two previous abortions at 12 weeks and 15 weeks. All investigations were done to find cause of abortions and were normal. On examination vision was reduced and fundus examination showed features of gyrate atrophy and confirmed by electroretinogram and serum ornithine levels. Gyrate atrophy is a rare genetic disorder characterized by chorioretinal atrophy. We advised genetic counseling for the patient. This case is presented as gyrate atrophy is rare in pregnancy and it may be associated with previous abortions. So fundus examination by an ophthalmologist was found to be important in finding out cause of abortion and this rare genetic condition.

Keywords: Gyrate atrophy, Serum ornithine, Electroretinogram, Pregnancy

INTRODUCTION

Gyrate atrophy is a very rare autosomal recessive disease due to pyridoxine dependent mitochondrial enzyme ornithine- γ -aminotransferase deficiency with Incidence less than 1/50000.

The retina shows extensive chorioretinal atrophy which is similar to gyri of the brain so hence the name gyrate atrophy. We report this case as gyrate atrophy is rare in pregnancy and may be cause of recurrent abortion.

CASE REPORT

34 years female came to ophthalmology outpatient department with complaints of defective vision for two years. History of amenorrhea for three months was present. She had two previous abortions at 12 weeks and 15 weeks. She was investigated for all autoimmune and connective tissue disorders and everything was normal. On examination vision right eye

6/60 not improving with pinhole and left eye 6/36, no improvement with pinhole. Eyelids conjunctiva, cornea was normal. Lens shows posterior subcapsular cataract and left eye more than right eye. Intraocular pressure was 18mmg by applanation tonometer. Fundus of both eyes shows extensive chorioretinal atrophy extending from periphery to centre sparing the macula (Fig. 1 and Fig. 2) extensive chorioretinal atrophy shows prominent vortex veins in periphery (Fig. 3). We suspected gyrate atrophy by above findings and we advised serum ornithine levels, visual fields and electroretinogram for this patient. Humphreys visual fields show 10° tubular fields. ERG for right eye shows non-recordable photopic&scotopic responses and left eye shows reduced and delayed combined response (Fig. 4). Serum Ornithine level was 320 μ M (normal—60-80 μ M) (Fig. 5). Patient was referred for genetic counseling and nutritionist for low protein diet. The patient was on follow up.

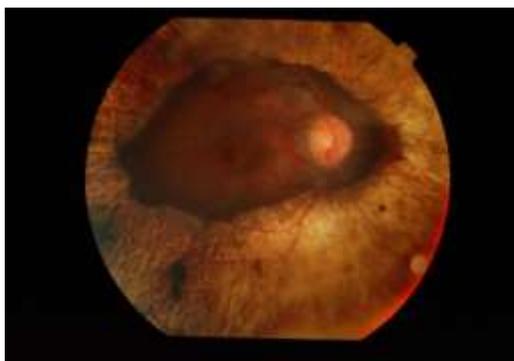


Fig. 1: Shows right eye extensive chorioretinal atrophy starting from periphery and towards central and sparing the macula with thinning of arteries and veins



Fig. 2: shows left eye extensive chorioretinal atrophy starting from periphery and spreading to central sparing the macula with thinning of arteries and veins

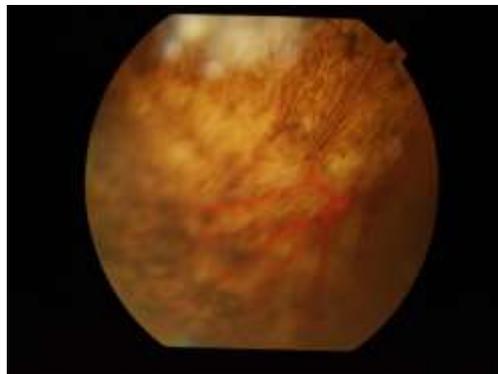


Fig. 3: Chorioretinal atrophy with visibility of vortex veins at periphery

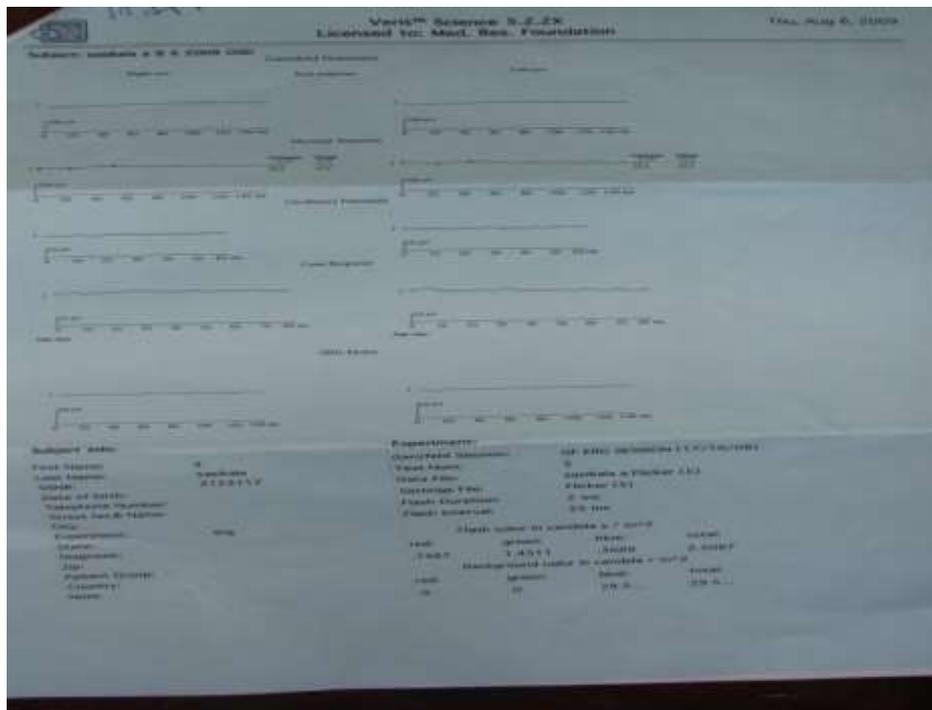


Fig. 4: Electoretinogram shows non recordable scotopic and photopic responses

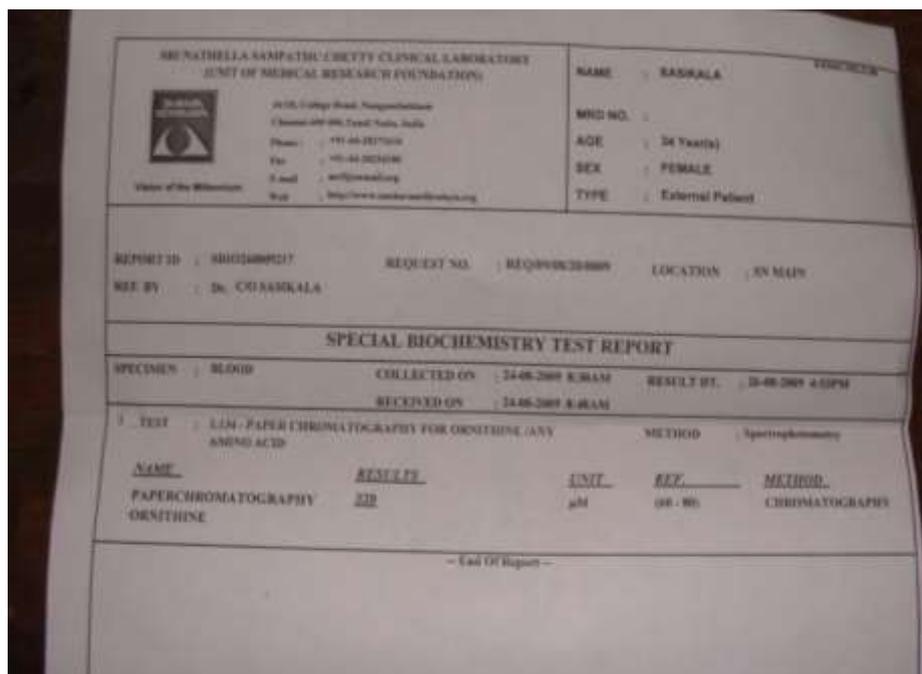


Fig. 5: Showing elevated serum ornithine levels

DISCUSSION

Jacobson described the first case of gyrate atrophy as atypical retinitis in 1888 [1]. Simell and Takki demonstrated the association of hyperornithinaemia and gyrate atrophy in 1973 [2]. Areas of chorioretinal atrophy coalesce and giving the fundus an appearance similar to cerebral gyri, hence this type of atrophy is called gyrate atrophy [3]. Gyrate atrophy is a rare autosomal recessive chorioretinal

atrophy due to pyridoxine dependent mitochondrial enzyme ornithine- γ -aminotransferase deficiency which was coded on chromosome 10q26 [4]. Most of the gyrate atrophy cases were reported from Finland, estimated at 1:50,000 [8].

In urea cycle ornithine was converted into glutamate and lysine by pyridoxine dependent enzyme ornithine- γ -aminotransferase.

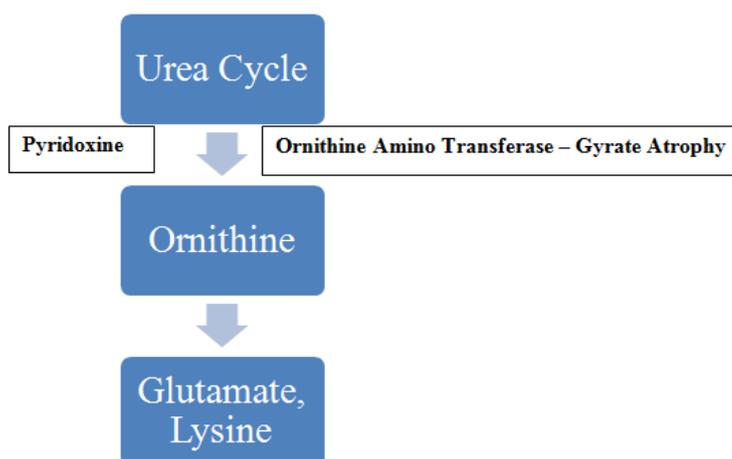


Fig. 6: Formation of glutamate and lysine by pyridoxine dependent enzyme ornithine- γ -aminotransferase

Deficiency of this enzyme cause 20-30 times increased levels of plasma ornithine level, which is toxic to Retinal pigment epithelium and choroid [5]. Axial myopia, posterior subcapsular cataract, night blindness are common ocular problems in gyrate atrophy. In fundus large geographic peripheral paving stone-like areas of atrophy of the RPE and

choriocapillaries in early stages. Gradual coalition of atrophic areas leads to form a scalloped border at the junction of normal and abnormal RPE in late stages. Fovea spares till later stage [6]. Non-recordable photopic & scotopic responses are seen in ERG. Biochemical changes are hyperornithenemia, hyperornithenuria, hypolysenemia. Pyridoxine

responders treated with Pyridoxine 300-500 mg daily to prevent the progression of the disease. Pyridoxine non responders are managed with low protein diet (10-15 g/day) and arginine restricted diet by avoiding dhal, soyabean, black gram, green gram [7].

REFERENCES

1. Jacobson E. Ein Fall von Retinitis Pigmentosa atypica. *Klin Monatsbl Augenheilkd*, 1888; 26: 202-206.
2. Simell O, Takki K; Raised plasma-ornithine and gyrate atrophy of the choroid and retina. *Lancet*, 1973; 1(7811): 1031-3.
3. Takkahashi O, Hayasaka S, Kiyosawa M, Mizuno K, Saito T, Tada K; Gyrate atrophy of choroid and retina complicated by vitreous haemorrhage. *Jpn J Ophthalmol.*, 1985; 29(2): 170-176.
4. Kanski JJ; Bowling B; *Clinical Ophthalmology: A systematic approach*. 7th edition, Saunders, 2011.
5. Heinänen K, Näntö-Salonen K, Leino L, Pulkki K, Heinonen O, Valle D *et al.*; Gyrate atrophy of the choroid and retina: lymphocyte ornithine-delta-aminotransferase activity in different mutations and carriers. *Pediatr Res.*, 1998; 44(3): 381-385.
6. Weleber RG, Kennaway NG; Clinical trial of vitamin B6 for gyrate atrophy of the choroid and retina. *Ophthalmology*, 1981; 88(4): 316-324.
7. Berson EL, Shih VE, Sullivan PL; Ocular findings in patients with gyrate atrophy on pyridoxine and low-protein, low-arginine diets. *Ophthalmology*, 1981; 88(4): 311-315.
8. Gyrate atrophy of choroid and retina. Available from <http://omim.org/entry/258870>