CASE REPORT

A rare case of Gyrate atrophy

Muhammad Waseem, Saquib Naeem, M Saleem Bajwa

Abstract

Gyrate atrophy is rarely seen in ophthalmological practice. We diagnosed one such case in our set-up. A middle aged male presented with decreased vision. Fundus showed patches of well-demarcated, scalloped atrophy of the pigment epithelium and choriocapillaris. Fundus fluorescein angiography revealed sharp contrast between normal and abnormal tissue which was typical characteristic of changes of Gyrate atrophy. Treatment includes pyridoxine (vitamin B6) and special diet. Prognosis usually remains poor and is a cause of disability. This case was being reported for general awareness.

Introduction

Gyrate atrophy is a rare entity. It is autosomal recessive bilateral disorder due to deficiency of enzyme Ornithine aminotransferase. Clinical features usually start in second decade. Symptoms include decreased midperipheral visual field leading to night vision problems, though central visual acuity is spared till late in the disease course. Fundus shows the gradual development of patches of well-demarcated, scalloped atrophy of the pigment epithelium and choriocapillaris in the midperiphery with characteristic border of surrounding hyperpigmentation, which eventually coalesce together till macula is involved resulting in decreased vision in later life. Fundus fluorescein angiography shows hyperfluorescence on the border of atrophic patch and sharp contrast between normal and abnormal tissue. Other ocular changes include mild cataractous opacities and mild maculopathy in later stages of the disease. Systemic features are mild which may include decreased intellect, myopathy and hair changes. Treatment includes pyridoxine (vitamin B_6) and special Arginine-free diet which may reduce the severity of the disease. Prognosis usually remains poor in later life[1-3].

Case report

A 50-year-old male presented in eye department PNS Shifa Karachi with complaint of decreased vision in right eye of 8 months duration. There was a history of a trauma to this eyeball of the same period. There was no other co-morbidity including night blindness.

✉ **Muhammad Waseem**

Prof and Head of Ophthalmology Department BUMDC & PNS Shifa Hospital, Karachi, Pakistan.
E-mail: muhammadwaseem57@gmail.com

Saquib Naeem
BUMDC & PNS Shifa Hospital, Karachi, Pakistan.

M Saleem Bajwa
BUMDC & PNS Shifa Hospital, Karachi, Pakistan.

Received October 17, 2011, Accepted November 09, 2011

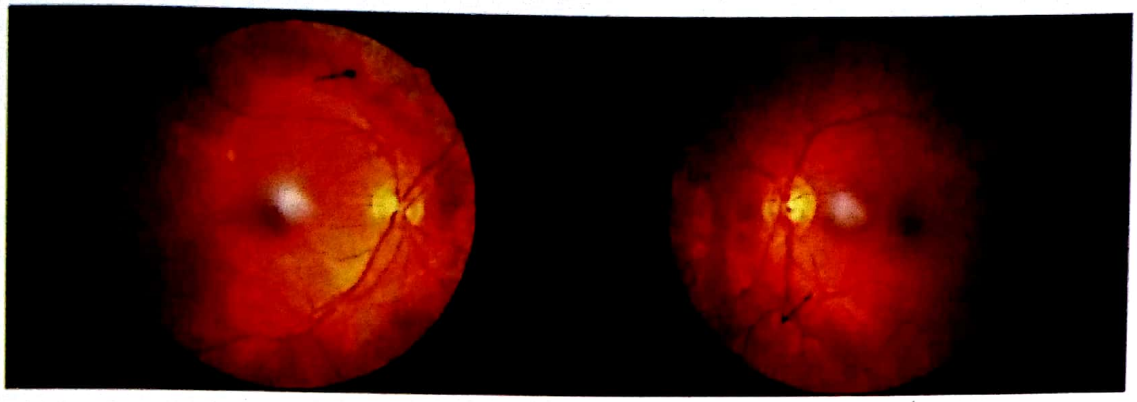
Vision in right eye was 6/9, correctable to 6/6 with glasses and the left eye was 6/6 unaided. Anterior segments were normal in both eyes. Pupils were reactive to light. Ocular motility was normal in all directions of gaze on both sides. The fundi oculi showed patches of well-demarcated, scalloped atrophy of the pigment epithelium and choriocapillaris with characteristic border of surrounding hyperpigmentation in mid-periphery. Optic disc and the macula were normal (Fig.1). Intraocular pressure was normal on both eyes. Systemic examination was normal.

On investigation, Fundus fluorescein angiography showed hyperfluorescence on the border of atrophic patch and a sharp contrast between normal and abnormal tissue (Fig.2) Patient was recommended tablet Pyredoxin 300 mg and to take Arginine- free diet. He was referred for visual fields, EOG and ERG and to the laboratory for enzyme assay.

Discussion

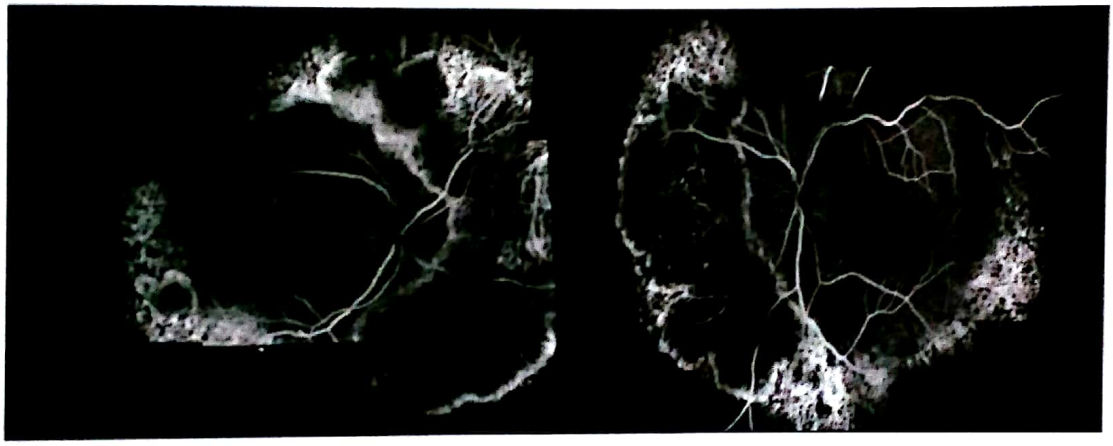
Gyrate atrophy, also known as essential atrophy of the choroid, occurs in early adult life is a rare condition[4]. So far, more than 150 cases have been identified[5]; approximately one third is from Finland). Ours will be 151th. Patient usually complains of defective night vision initially but somehow, this was not the presenting complaint in this case. He attributed his defective vision to the trauma to his right. The characteristic fundus findings and fluorecein angiographic pictures so typical of Gyrate atrophy were all present in our patient. Systemic involvement can occur but there was no such evidence in our patient. The treatment of choice is vitamin B6 and low Arginine diet but it is yet to be shown that modification of the metabolic control has any influence on the eye disease[6-7] .We have started the treatment but so far we have not got the feedback and so the response could not be judged.

If started at an early age, long-term substantial reduction of plasma ornithine levels may appreciably slow the progression of the chorioretinal lesions[8]. We would like to follow this case at six months interval.



Right Eye Left Eye

Figure-1Fundus photograph of the patient



Right Eye Left Eye

Figure-2 Fundus Fluorescein Angiographs of the patient

Conclusion

Gyrate atrophy of the choroid and retina is a rare inborn metabolic disorder whose ocular effects may occur at any time during the course of the disease. It should be suspected in any patient who presents with subtle ocular symptoms in presence of chorioretinal atrophic patches in mid-periphery and can be confirmed on fundus fluorescein angiography and plasma Ornithine levels. Result of the treatment is fruitful if detected and treated early in life.

References

1. Juan Jr E, Noorily SW, Townsend-Pico WP, Chern KC. Hereditary Disorders in Childer and Young Adults. Ch 57.In: Wright K W, ed. Textbook of Ophthalmology, Williams & Wilkins 1997; 812-3.

2. Kanski JJ. Clinical Ophthalmology: A systematic approach, 4th ed. oxford: Boston; 1999.

3. Epstein RL. Hyperornithinemia: Gyrate Atrophy of the Choroid and retina. Inborn Metabolic Disorders and the Eye.Ch 28 In: Peymann GA, Sanders DR, Goldberg MF. eds. Pricipal and Practice of Ophthalmology. Vol 3, WB Saunders Company 1980;1759-60.

4. Miller SJH. Diseases of the Uveal Tract.Ch 18. Parsons' Diseases of the Eye. 17th ed. Churchill Livingstone 1984; 167.

5. Gyrate atrophy of the choroid and retina. Available at: http://ghr.nlm.nih.gov/condition/gyrate-atrophy-of-the-choroid-and-retina, 2009.

6. Newell FW. Gyrate Atrophy of the Choroid. The Middle Coat:The Uvea Ch 15. In: Ophthalmology Prinicipal and Concepts. 6th ed. The C.V. Mosby Company 1986; 287.

7. Bird AC. Retinal Receptor Dystrophies.In:Miller SS. Clinical Ophthalmology, Wright 1987; 208-9.

8. Kaiser-Kupfer MI, Caruso RC, Valle D. Gyrate atrophy of the choroid and retina: further experience with long-term reduction of ornithine levels in children. Arch Ophthalmol. 2002;120:146-53.