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CASE REPORT

CKD or is it a Fabry Disease: a case report

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ABSTRACT

Fabry disease is a rare X-linked lysosomal storage disorder caused by mutations in the GLA gene, leading to deficient activity of the enzyme alpha-galactosidase A. This case report presents a rare instance of Fabry disease in a 37-year-old female patient, highlighting the unique clinical presentation with multisystem involvement. The patient presented with a complex array of symptoms, including loss of appetite, generalized body weakness and significant blurring of vision accompanied by a history of hypertension and chronic kidney disease (CKD). Laboratory investigations revealed deranged complete blood count, elevated renal function parameters, and significant proteinuria. Ophthalmic evaluation showed decreased visual acuity in right eye with normal visual acuity in left eye, typical whorl like deposits in both corneas, tortuous conjunctival vessels and tortuous retinal vessels in both retinas with venous beading and flame shaped hemorrhages prominent in left eye whereas right macula had significant macular edema. On further screening on her offspring, her male child also had similar whorl like deposits in both corneas. This case is a rare and unique example of Fabry disease in a female patient, with symptoms affecting multiple organ systems, including the renal, cardiovascular system and eye. It underscores the importance of maintaining a high index of suspicion for Fabry disease, even in female patients, and the need for a comprehensive diagnostic approach to ensure timely diagnosis and appropriate management. Early recognition of this rare condition in females is crucial for the implementation of targeted therapies to prevent the progression of multi-organ damage.

INTRODUCTION

Fabry disease (systemic angiokeratoma) is an X-linked recessive inherited lysosomal disorder due to α-galactosidase, an enzyme deficiency in the body caused by mutations in the GLA gene. The progressive accumulation of glycosphingolipids, specially globotriaosylceramide (GL-3), in the lysosomes of the cells of multiple tissues and organs causes progressive impairment of renal and cardiac functions as well as cerebrovascular and ocular pathologies. The estimated prevalence of Fabry's disease is about one per 80,000 to 117,000.[1] Due to X-linkage, it affects both males and females. Males usually have an earlier onset and a more severe form of the disease whereas female carriers are asymptomatic.[2] Age of onset can range from early childhood to the 7th decade with mean age 45±17 years (range: 22-75 years).[3]

CASE REPORT

A 37 years female from Bullingtar, Chitwan was referred to Department of Ophthalmology from Department of Nephrology ward of Chitwan Medical College with the provisional diagnosis of chronic kidney disease with hypertension for routine ophthalmic evaluation to look for hypertensive retinopathy changes. She had presented with the complaint of loss of appetite for 3 months and weakness of body for 5-6 days and there was history of significant blurring of vision in both eyes (right more than left) for 2 weeks. She had no past medication history. She was non-smoker, non- alcoholic and had no history of substance abuse. On general examination she was conscious, well oriented to time, place and person, looked mildly pale. Her vitals were stable except blood pressure being 160/100 mm of Hg despite anti-hypertensives. Her general systemic examinations were insignificant and were within normal limits.

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Her ophthalmological evaluation was done and found to have uncorrected visual acuity (VA) with Snellen chart of 6/24 on her right eye and with pin hole improvement to 6/18 and 6/6 partial in left eye which did not improve with pin hole. Intraocular pressure (IOP) was 17 mmHg in both eyes with Goldmann applanation tonometry. Slit lamp bio-microscopy revealed increased tortuosity in conjunctival vessels. There was a whorl like deposits in corneal epithelium bilaterally (cornea verticillata) (Fig. 1).

Anterior chamber was quiet with normal depth. Lens was clear with no any significant opacities. Posterior segment evaluation revealed vertically oval optic disc with normal margins and dimensions in both eyes. Disc was slightly pale bilaterally. There was mild vessel tortuosity and venous beading in both eye with few flame-shaped hemorrhages present only in the left eye in infero-nasal quadrant. There was significant macular edema on the right eye which justifies the decrease in visual acuity in right eye which wasn't improved with refraction. These findings were documented in Amsler-Dubois chart (Fig. 2). FAF imaging couldn't be performed due to unavailability of resource. Further ophthalmological evaluation by OCT, Visual field and Fundus photo couldn't be performed due to poor resource availability.

On further elaboration of history, she has a family history of blindness in her grandfather, sudden death of her sibling from unknown cause at young age. On family screening for Cornea verticillata, her siblings didn't have significant ocular findings, but her elder son has similar corneal verticillata (Fig. 3). Her elder son's visual acuity and fundus examinations are all within normal limit. Her echocardiography showed mild MR, Mild TR, dilated LA and LVEF=60%, rest within normal limit.

On her ultrasonographic scan of abdominal and pelvic there was increased cortical echogenicity in both kidneys with poorly maintained cortico-medullary differentiation (CMD), suggestive of stage V CKD. Her blood parameters was deranged with Hemoglobin 8.7gm%, platelets 142,000/mm^3, TLC 7,120/mm^3, RBS 111mg/dl, Serum uric acid 11.47mg/dl, serum creatinine 12.64mg/dl, blood urea 236mg/dl, serum potassium (K+) 3.74mmol/l, serum sodium (Na+) 142mmol/dl, ANA (IFA) positive reaction at 1:40, viral markers (HIV, HBSAG, ANTI-HCV) –Non reactive, Urinary protein 204.7mg/dl, Urinary creatinine 32.53mg/dl, protein/ creatinine ratio (PCR) 6292.652mg/g. Urine R/E RBC+, Epithelial cells (2-4), Pus cells (2-4), Glucose (Nil), Bilirubin (Nil), pH acidic. In addition, CT head showed normal findings and blood culture showed no growth in 7 days.

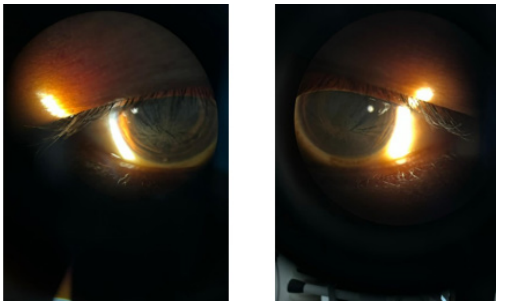

Figure 1: Cornea verticillata in patient with Fabry Disease

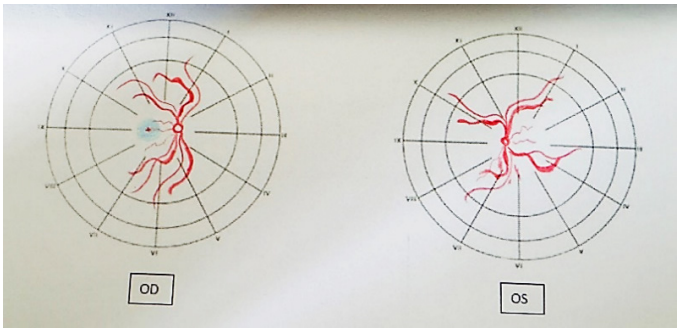

Figure 2: Fundus drawing representing the posterior segment findings

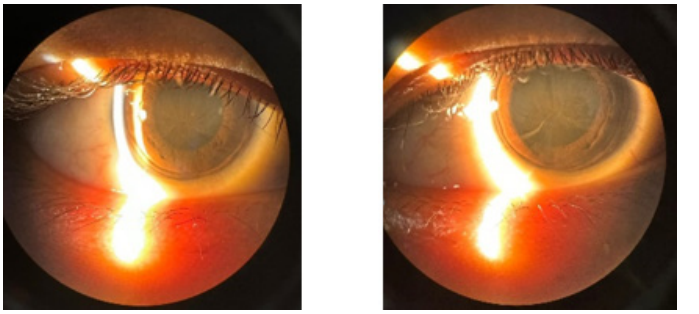

Figure 3: Cornea verticillata in patient's asymptomatic male child

Figure 4: Pedigree chart showing familial trait

DISCUSSION

Fabry disease (systemic angiokeratoma) is an X-linked inherited lysosomal disorder due to α-galactosidase, an enzyme deficiency in the body caused by mutations in the GLA gene. The symptoms of Fabry disease are mainly due to the tissue accumulation of globotriaosylceramide (GL-3), which may include a burning sensation in the hands and feet, reduced sweating, nausea, abdominal pain, postprandial diarrhea, and developmental disorders[4]. Conjunctival and retinal vessel tortuosity, and corneal verticillata are frequently observed in Fabry disease[5]. Cornea verticillata is the most common and earlier ocular manifestation of Fabry disease, detection of which should prompt consideration of Fabry disease as a provisional diagnosis. The patient should be examined for the extra-ocular findings such as progressive cardiomyopathy, nephropathy, renal failure, and cerebrovascular events. The rare findings are retinal artery vein occlusions, anterior ischemic optic neuropathy, optic disc pale or atrophy, disc edema and tear film disorders[2].

Fabry disease being an X- linked condition family screening is crucial, as early detection of this disease may help in treating the patient with the disease and thus decrease morbidity

and mortality[6]. Although the overall prevalence of FD is low in patients with kidney involvement, screening, especially in patients who have not yet undergone kidney replacement therapy, is important, in order to provide timely and effective treatment interventions, including disease modifying therapies[7]. With the advent of enzyme replacement therapy, it is important that general practitioners and physicians in a range of specialties recognize the signs and symptoms of Fabry disease so that effective treatment can be given[8].

CONCLUSION

Cornea verticillata, dilated tortuous conjunctival and retinal vessels are the most common and earlier ocular manifestation of Fabry disease and thus, patient with unexplained CKD must be referred for Ophthalmological evaluation for early screening of Fabry disease.

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