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Childhood Ocular Cystinosis- A Case Report

Zubaida Noor*[1], Sujit Kumar Biswas[2], Nuruddin Zahed[1]
[1] Resident, Chittagong Eye Infirmary & Training Complex
[2] Consultant, Chittagong Eye Infirmary & Training Complex

*Corresponding Author: Zubaida Noor

Abstract:-
Aim: To present a rare case of childhood ocular cystinosis.
Case Report: We report a case of 10 years old girl with ocular cystinosis presented with the complain of photophobia, watering, sometimes redness and foreign body sensation. On ophthalmic examination visual acuity was 6/6 in both eyes and slit-lamp examination reveals fine cystine crystal deposition in both corneal. Rest of the ocular examination was unremarkable.
Conclusion: Cystinosis is a rare Autosomal recessive disorder which is basically characterised by deposition of amino acid cystin within lysosome that damages various organs and tissues mainly kidney and eyes. A multidisciplinary approach icluding ophthalmologist, nephrologist can manage a case of cystinosis.

Keywords:- *Cystine Crystals, Corneal Deposists, Photophobia.*

I. INTRODUCTION

Cystinosis is a rare form of genetic disorder that is Autosomal recessive lysosomal storage disorder with an incidence of 1 case per 1,00,000-2,00,000 live births[1]. It was first published in 1903 by Swiss biochemist and physiologist Emil Abderhalden (1877-1950) [2]. Cystinosis causes deposit of cystin in various organ including eyes and kidneys. Ocular cystinosis causes photophobia, watering, visual problem and mild discomfort and there is risk of glaucoma. Early detection is needed to prevent these complications mainly to prevent visual impairment.

II. CASE REPORT

A 10 years old girl came to cornea outpatient department with her mother with complain of severe photophobia, sometimes redness and foreign body sensation. On examination her visual acuity was 6/6 in both eyes. Slit-lamp examination showed diffuse stromal crystals deposit over entire cornea of both eyes (Fig. 1).

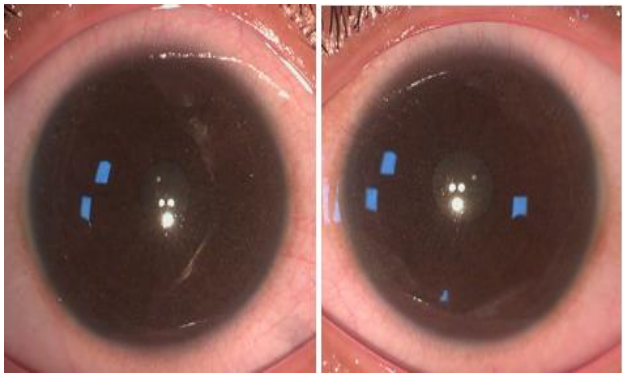

Fig 1: Fine crystals deposited all over both of the cornea

Other anterior segment examination and fundus was normal. The other family members did not have similar findings. The patient was advised to do some systemic examinations including kidney-function test, liver-function test, thyroid-function test. Serum bilirubin was 0.5 mg/dL, serum SGOT 30 µ/L, serum ALP 180 U/L, serum ures 19.5 mg/dl, serum BUN 9.1 mg/dl, serum creatinine 0.5 mg/dl with eGFR 100.7/ml/min/1.73m2, free T3 2.85pg/ml, free T4 1.20 ng/dl and TSH was 1.67µIU/ml. USG of whole abdomen was normal (Fig. 2).

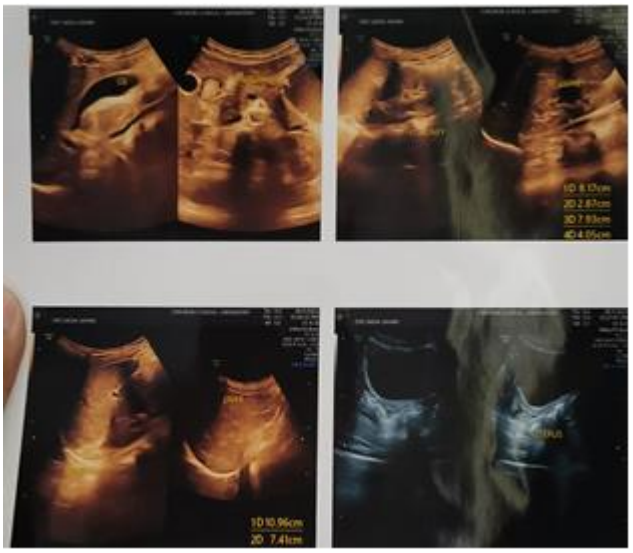

Fig 2: USG of whole abdomen.

Systemic examination revealed no abnormality. We did color fundus photograph os both eyes and showed completely normal and there was no cysteine crystal deposition in the retina (Fig. 3).

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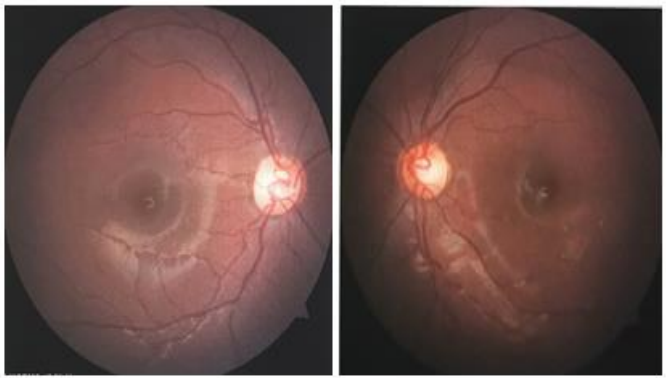

Fig 3: Fundus photos.

On the basis of ocular and systemic examination she was diagnosed as a case of ocular (non-nephrotic) cystinosis. The patient was advised to use photochromic spectacles to prevent photophobia and artificial tear eye drop for foreign body sensation. But was unable to get cysteamine eye drops due to lack of preparation in our country. She is advised to follow-up regularly for corneal stability checkup.

III. DISCUSSION

Ocular cystinosis is rare genetic disorder that affects both male and female equally, commonly diagnosed in childhood between the age of 1 to 7 years. Mostly seen in European descent ,but also has been reported in people of all ethnic background. It occurs mainly from mutations in the gene CTNS [3] mapped on chromosome 17p13.2 encoding the protein cystinosin,the lysosomal cystine transporter [4]. This mutation cause dfective cystine transport out of lysosome that leads to excessive accumulation and crystalization in various tissues ,specifically in kidneys and eyes, later on thyroid, liver, testes, pancrease, muscles, brain, bone marrow and spleen causing different symptoms [1,3]. There are 3 types of cystinosis, the most severe one is Infantile nephrotic cystionosis, less severe is Juvenile nephrotic cystenosis and extremely rare is Ocular non nephrotic cystionsis.

In Ocular cystinosis there is no systemic manifestations, only shows corneal deposits and usually diagnosed in adulthood [5]. It mainly causes photophobia, decreased visual acuity and mild discomfort. Other than cornea it can affect other ocular structures such as conjunctiva, anterior chamber, lens and retina. Conjunctival involvement can cause irritation, redness and mild discharge [6]. Involvement of retina is rare but if so can cause pigmentary retinopathy with visual field deficit. Cysteamine eye drops hault the progression of retinal degeneration and can prevent vision loss [5,6].

Regular ophthalmic examination including visual assesment, slitlamp biomicroscopy, angle evaluation, ocular investigation such as Colour fundus photograph, anterior segment OCT, in vivo confocal microscopy (IVCM) can prevent visual loss and other complications. IVCM is the Gold standard method of observing corneal cystine deposition [7]. The treatment for Cystinosis is oral an eyedrop based cysteamine both are approved by FDA. This medicine is referred as “orphan medicines” as they are used in rare diseases. Cysteamine drop helps by dissolving cystine crystals in tissues and give relief from photophobia and discomfort. Study shows 0.55% cysteamine hydrochloride drops are well tolerated and reduces corneal deposits and improve visual acuity in a patient of ocular cystinosis [8]. If cystine accumulation aggravated involving limbal stem cell deficiency and peripheral corneal vascularization in addition with cysteamine therapy, corneal transplantation such as Penetrating keratoplasty is performed. But the success of corneal transplantation is limited due to recurrent cystine deposition in donor cornea [9].

Overall Ocular cystinosis management requires a multidisciplinary approach. Further research is needed to prevent or reduce complication of this disease and for newer treatment modalities.

IV. CONCLUSION

Visual handicap is the worst complication of cystinosis that need to be detected early to prevent such complication. Good monitoring and regular follow-up by both ophthalmologist and nephrologist is needed .They should be aware of all the complication related to the disease and work closely to ensure timely intervention and optimal outcomes.

FINANCIAL INTEREST – Nil

CONFLICTS OF INTEREST- there are no conflicts of interest.

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