Original Research Paper

Volume-9 | Issue-12 | December - 2019 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Ophthalmology

A CASE OF CORNEAL CYSTINOSIS IN A PATIENT WITH CHRONIC RENAL FAILURE

Dr. Nikita Goel	3rd Year Resident, Department of Ophthalmology, B.J. Medical College, Civil Hospital, Ahmedabad.
Dr. Sahil Sharma*	2nd Year Resident, Department of Ophthalmology, B.J. Medical College, Civil Hospital, Ahmedabad. *Corresponding Author
Dr. Rohit Dureja	MS, Department of Ophthalmology, B.J. Medical College, Civil Hospital, Ahmedabad.
Dr. Tanvi Choudhary	3rd Year Resident, Department of Ophthalmology, B.J. Medical College, Civil Hospital, Ahmedabad.

ABSTRACT Cystinosis is a rare autosomal recessive disorder characterized by the intracellular accumulation of disulphide of the aminoacid cysteine in various organs and tissues. Infantile nephropathic cystinosis is the most severe type. Although renal disease is the most prominent feature of cystinosis the corneal cysteine crystal formation remains a major complication leading to photophobia, corneal erosion and keratopathy. Corneal crystal accumulation and pigmentary retinopathy were originally the most commonly described ophthalmic manifestation but successful kidney transplantation significantly changed the natural history of the disease. A case of a 9 year old boy diagnosed with nephrotic cystinosis was reported. He developed renal proximal tubulopathy (or fanconi syndrome) at 7 months of age and then uprogressed to end stage renal disease. He presented with severe photophobia and decreased visual acuity. Ocular cystinosis was diagnosed on observing the typical crystals in cornea. OCT showed multiple areas of stromal hyperreflectivity due to crystal deposits within the cornea.

KEYWORDS :

CASE REPORT

A 9 year old boy was referred to us from Nephrology department for ocular complaints of severe photophobia and decreased visual acuity in both eyes. He was diagnosed initially at the age of 7 months with proximal tubulopathy and has now progressed to end stage renal disease. His investigations revealed-low potassium (2.1mEq/L), low sodium (130mEq/L) and phosphate levels of 1.9mg/dL. Urine analysis showed glycosuria (sugar 2+), albuminuria (albumin 2+) consistent with fanconi's syndrome. CT scan showed moderately enlarged kidney.

The visual acuity was counting finger four metres in both eyes. The intraocular pressure could not be measured because patient was uncooperative.Slit lamp examination revealed moderate corneal edema and multiple fine shiny crystal like deposits in the corneal epithelium and stroma in both eyes. We evaluated the status of cornea using anterior segment OCT which showed hyperreflectivity in the stroma in both eyes. Cystagon (cystamine bitartrate) eye drops were prescribed but due to unavailability of the eye drops he was given an option of surgery in the form of penetrating keratoplasty.

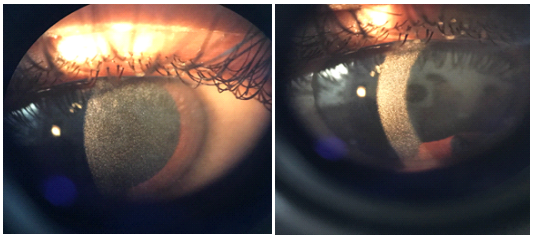

Figure 1 & 2 : Corneal cysteine deposits seen as fine crystals in the corneal stroma one slit lamp examination

DISCUSSION

Cystinosis is a rare autosomal recessive metabolic disorder characterized by defect in lysosomal cysteine transporter, Cystinosin(CTNS). It is mapped to CTNS gene on short arm of chromosome 17 which encodes cystinosin a467 amino acid(AA) transmembrane protein. The CTNS gene consists of 12 exons. Various mutations have been identified thoughout the gene and the most common one is a large 57kb deletion which removes the first 9 exons and most of the 10th exon. The main pathological finding is intralysosomal accumulation of cysteine crystals in various tissues such as conjunctiva, cornea, kidney basement membrane and associated organ dysfunction. The corneal accumulation of needle shaped cysteine crystals is characteristic and progresses from epithelium towards endothelium through stroma.

Cystinosis is the first described lysosomal storage disorder for which a safe and efficacious treatment exists. There are three main phenotypes of cystinosis- 1. Infantile nephropathic cystinosis- classic form, most common(95%) and most severe. 2. Juvenile-less severe, late onset. 3. Adult cystinosis- benign non nephropathic form. Definitive diagnosis is achieved by quantitative estimation of leucocyte cysteine level which is not routinely available. Treatment consists of cysteine depleting therapy in addition to management of renal tubular acidosis. The typical untreated child with cystinosis has short stature, rickets, photophobia.

Our patient had signs of rickets and growth failure. The corneal findings were confirmed on optical coherence tomography. Topical cysteamine treatment has been proven to be a safe and efficient way of dissolving corneal crystals in cystinosis patients. It also relieves the symptoms of photophobia and blepharospasm. We prescrived topical cystamine drops to our patient but it is not easily available therefore patient was given an option of optical penetrating kerotaplasty.

Figure 3 : The child showing malnutrition with renal rickets

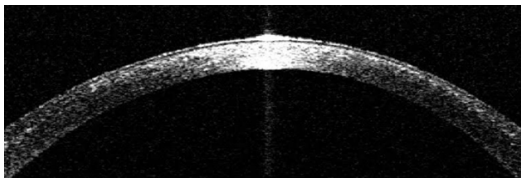

Figure 4 : Corneal cysteine deposition seen as hypersensitivity in the stroma on anterior segment optical coherence tomography

CONCLUSION

Ocular involvement has been shown to be present in all forms of

disease .Examination for corneal cysteine crystals is a simple and specific diagnostic test and has proved very useful in diagnosing cystinosis. Extent of corneal involvement may serve to reflect the course and severity of systemic disease. The most effective treatment for corneal cystinosis is topical cysteamine eye drops (a cysteine depleting drug)but due to logistic difficulty of procurement an alternate option of surgical intervention can be given which holds good results.

REFERENCES

1. Nesterova G, Gahl WA. Cystinosis: The evolution of a treatable disease. Pediatr Nephrol 2013;28:51-9. Back to cited text no. 1
2. Burki E. Über die Cystinkrankheit im Kleinkindesalter unter besonderer Berücksichtigung des Augenbefundes. Ophthalmologica 1941;101:257-72. Back to cited text no.
3. De Graaf-Hess A, Trijbels F, Blom H. New method for determining cystine in leukocytes and fibroblasts. Clin Chem 1999;45:2224-8. Back to cited text no. 3
4. Thoene JG, Oshima RG, Crawhall JC, Olson DL, Sghneder JA. Intracellular cystine depletion by aminothiols in vitro and in vivo. J Clin Invest 1976;58:180-9. Back to cited text no. 4
5. Lyseng-Williamson K. Cystadrops® (cysteamine hydrochloride 0.55% viscous eye-drops solution) in treating corneal cystine crystal deposits in patients with cystinosis: a profile of its use. Drugs Ther Perspect 2017;33:195. Back to cited text no. 5
6. US-FDA access data. retrieved on 10 October 2018 from; <https://www.accessdata.fda.gov/drugsatfda_docs/nda/2012/200740Orig1s000MedR.pdf>. Back to cited text no. 6