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Rare Case Report: Cystinosis

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Abstract: ***A 16 year old female presented to eye OPD on 22/12/2021 at S.S.G. Hospital, Vadodara with chief complaints of dimness of vision and severe photophobia in both eyes. Patient is a diagnosed case of Nephropathic Cystinosis. She has hypophosphatemic rickets, Hypothyroidism, metabolic acidosis, growth stunting and ocular involvement since 2013. She is on systemic and topical treatment for eye. (Cysteamine capsule and eye drops) Slit lamp examination revealed corneal and conjunctival crystalline deposits. Dilated fundus examination shows OU Disc pallor and peripheral crystalline retinopathy. Patient had genetic test report showing mutation in the gene CTNS, located on chromosome 17, which codes for cystinosin, the lysosomal cystine transporter.***

Keywords: Cystinosis, Cysteamine Eye Drops, CTNS Gene

1. Introduction

Cystinosis is a rare, multisystem genetic disorder characterized by the accumulation of an amino acid called cystine in different tissues and organs of the body including the kidneys, eyes, muscles, liver, pancreas and brain. Cystinosis was first described in the medical literature in 1903 by Abderhalden.[1] It is an autosomal recessive disorder resulting from accumulation of free cystine in lysosomes, eventually leading to intracellular crystal formation throughout the body. Defective function of the lysosomal membrane protein cystinosin, results from mutations of the cystinosis gene (CTNS). That resides on chromosome 17p13.[2] Cystinosis affects approximately 1 in 100,000 to 200,000 newborns and there are only around 2,000 known individuals with cystinosis in the world. It is often considered a disease of fair-skinned individuals of European descent; however, it is known to occur in blacks, Hispanics, and other races around the globe. The male-to-female ratio among cystinotic children has been reported to be 1.4:1.[3]

2. Case Report

A 16 year old Indian female patient presented to eye OPD on 22/12/2021 at S.S.G. Hospital, Vadodara with chief complaints of dimness of vision and severe photophobia in both eyes. Patient is a diagnosed case of Nephropathic Cystinosis with ocular involvement since 2013. Visual acuity OU was 6/18 and (not corrected further by glasses) slit lamp examination revealed corneal and conjunctival crystalline deposits. Dilated fundus examination showed OU Disc pallor and peripheral crystalline retinopathy. IOP (tonopen) was within normal limit. VEP OU showed a normal latency and amplitude in both eyes. ERG OU showed a delay in latency and reduced amplitude in right eye. Patient had genetic test report showing mutation in the gene CTNS, located on chromosome 17, which codes for cystinosin, the lysosomal cystine transporter. She has hypophosphatemic rickets, Hypothyroidism, metabolic acidosis, growth stunting. She is on systemic and topical treatment for eye. (Cysteamine capsule and eye drops)

3. Results

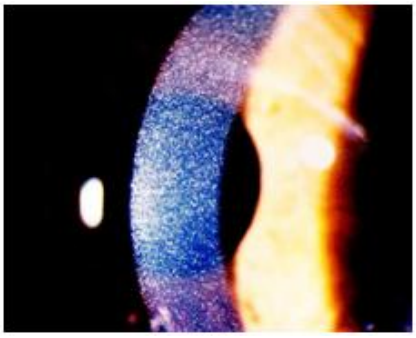

Figure 1: Slit lamp examination: Crystalline deposits in corneal storma

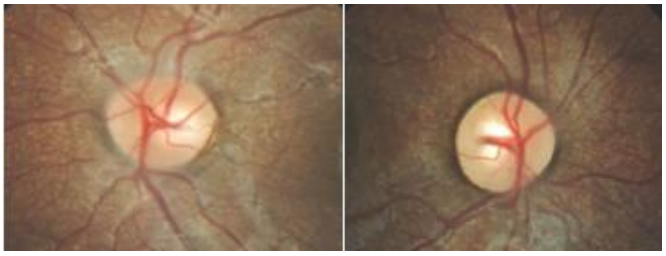

Figure 2: Fundus photographs waxydisc pallor and crystalline retinopathy

4. Discussion

A 16 yrs old Indian female patient, diagnosed case of Nephropathic Cystinosis has a short stature, growth stunting, faircomplexion, skeletal dysplasia. Crystalline deposits in corneal stroma, ground glass conjunctiva and peripheral retinal hypopigmentation and mottling of the retinal pigment epithelium was seen. Topical cysteamine eye drops are efficient at dissolving corneal crystal and relieving the symptoms of photophobia, blepharospasm and ocular pain. Cysteamine eye drops 2 hourly have helped to relieve ocular pain and photophobia by decreasing corneal crystal deposition. Drops are needed to be kept cool and used within a week of opening.

5. Conclusion

There are three distinct types of cystinosis each with slightly different symptoms: nephropathic cystinosis, intermediate cystinosis, and non-nephropathic or ocular cystinosis.[4]

Infants affected by nephropathic cystinosis initially exhibit poor growth and particular kidney problems (sometimes called renal Fanconi syndrome). The kidney problems lead to the loss of important minerals, salts, fluids, and other nutrients. The loss of nutrients not only impairs growth, but may result in soft, bowed bones (hypophosphatemic rickets), especially in the legs. The nutrient imbalances in the body lead to increased urination, thirst, dehydration, and abnormally acidic blood (acidosis). By about age two, cystine crystals may also be present in the cornea. Without treatment, children with cystinosis are likely to experience complete kidney failure by about age ten. With treatment this may be delayed into the patient's teens or 20s. Other signs and symptoms that may occur in patients include muscle deterioration, blindness, inability to swallow, impaired sweating, decreased hair and skin pigmentation, diabetes, and thyroid and nervous systemproblems.

Intermediate cystinosis typically begins to affect individuals around age twelve to fifteen. Malfunctioning kidneys and corneal crystals are the main initial features of this disorder.[5]

People with non-nephropathic cystinosis do not show experience growth impairment or kidney malfunction. The only symptom is photophobia due to cystine crystals in the cornea.[6]

Conflicts of Interest: Declared, authors have no conflict of interest

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