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The first confirmed case of cystinosis in children in the Republic of Kazakhstan.

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Case Study : Background: Cystinosis is a lysosomal storage disease characterized by an intracellular accumulation of cystine in different organs and tissues, leading to potentially severe organ dysfunction. Three forms of cystinosis have been described primarily based on age of presentation and severity of phenotype: 1) Infantile (nephropathic) form, 2) Late-onset (juvenile) form, 3) Adult (benign) form. It is an autosomal recessive lysosomal storage disorder caused by mutations in the CTNS gene encoding for the carrier protein cystinosin, transporting cystine out of the lysosomal compartment. Infantile cystinosis, also referred to as nephropathic cystinosis, is the most common form of cystinosis and is estimated to affect 1 of every 100,000 to 200,000 children. Case description: Patient S. is a 14 years old male presenting with the signs valgus deformity of the lower extremities and chronic kidney disease stage G2: creatinine 80.30-90.3 mmol/l, GFR 72.73-64.67 ml/min, urea 4.20mmol, proteinuria 4.29 g/l, Cr/Pr: 564 mg/ml. During hospitalization the patient was found to have genetic study: Gene: CTNS, Position (GRCh37/hg19): chr17:G.3559990C>G, chr17:g.3561395del18, Genotype: heterozygote, Exon: 9, 10, cDNA variant (AK): c.582C>G (p.Tyr194Ter), c.779_796del (p.Thr260_Leu266delinsIle), Allele frequency: n/a, Reference sequence: NM_001031681.2, Reading depth: 470x, 48x. Examined by an ophthalmologist: OI Corneal cystinosis. Mild myopia with astigmatism. Result: It is the first case of Cystinosis, juvenile form reported in Kazakhstan. Assigned symptomatic and pathogenetic therapy observed in dynamics. Conclusion: According to international recommendations, the therapeutic tactics of this disease are symptomatic and pathogenetic therapy. Pathogenetic drugs are Cysteamine topical eye drops and Cysteine (cysteamine) capsules are taken orally. Due to the rarity of the disease, current treatment is not possible in the Republic of Kazakhstan. In this regard, the preparation of a protocol of examination and treatment according to the international protocol has been initiated.

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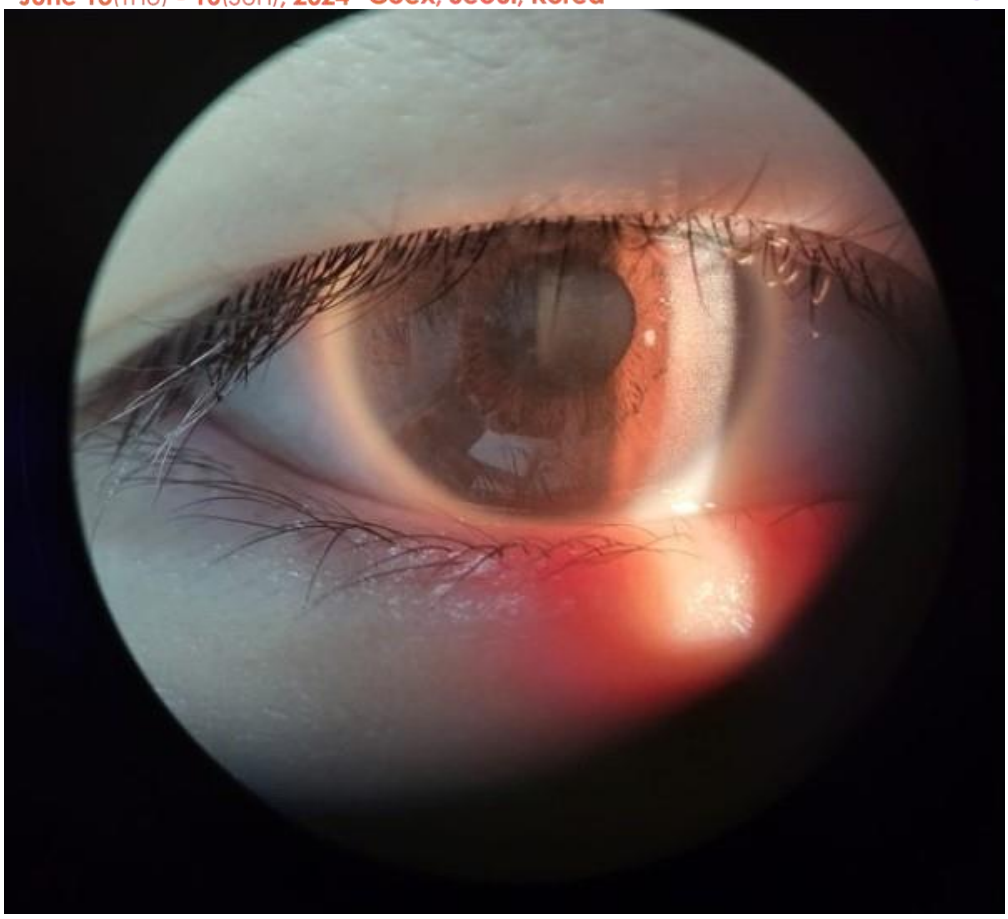

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Completed Scans
Right / OD
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Pachymetry
Cornea Line