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Detection of 57-kb Deletion of the *CTNS* Gene in Patients with Nephropathic Cystinosis from West Azerbaijan Province of Iran

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ABSTRACT

Introduction: Cystinosis is a rare autosomal recessive lysosomal storage disease caused by mutations in the CTNS gene, resulting in the absence of the cystinosin transporter. This deficiency leads to the abnormal accumulation of cystine within lysosomes, affecting body tissues and organs. Early detection of elevated cystine level in white blood cells is crucial. The ethnicity of the tested population greatly influences the genetic base of cystinosis. The most public pathogenic *CTNS* mutations in Northern Europe and North America is a large deletion of the *CTNS* gene (57-kb del). This mutation affects the promoter and the first 10 exons of the CTNS gene, as well as CARKL and TRPV1 genes. The 57-kb deletion of the *CTNS* gene is found in more than 50% of mutant alleles in Northern Europe and North America. Conversely, the frequency of this mutation is low in Middle Eastern nations. This study aimed to detect the 57-kb deletion of the *CTNS* gene in patients with nephropathic cystinosis from West Azerbaijan province of Iran.

Methods and Materials: Thirteen patients with nephropathic cystinosis were involved in the study. An ophthalmologist and pediatric nephrologist confirmed cystinosis diagnosis. After obtaining informed consent from the patient's parents, 3-4 ml of whole blood taken from the patients was transferred to 500 µl EDTA-containing tube. DNA was isolated by salting out procedure from whole blood of the tested patients. Exons 3 to 10 of the *CTNS* gene were amplified with specific primers. Afterwards, PCR products were directly sequenced by an ABI 3700XL Genetic Analyzer. The sequences were analyzed by CHROMAS software.

Results: The 57-kb deletion of the *CTNS* gene was not found in patients with nephropathic cystinosis.

Conclusion and Discussion: Our report contribute to the understanding of the genetic basis of nephropathic cystinosis in the West Azerbaijan Province of Iran. It paves the way for family counseling, carrier screening, and prenatal diagnosis in the north-west region of Iran. The findings of this study are valuable for health systems and health-related policy organizations, enabling them to undertake necessary actions to prevent fatal disease. The pathogenic 57-kb deletion mutation of the *CTNS* gene was not found in the tested population in our study. Evaluating other common mutations are suggested.

Keywords: Cystinosis, Iran, Mutation