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Frequency of Pathogenic Mutation c.971-12G>A in Intron 11 of *CTNS* gene in Nephropathic Cystinosis Patients Referred to Shahid Motahari Educational-Therapeutic Hospital

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ABSTRACT

Introduction: Cystinosis is a rare autosomal recessive disorder caused by the intra-lysosomal accumulation of cystine in tissues due to mutations in the *CTNS* gene. This disease has been known clinically for more than 100 years. As of 2016, more than a hundred mutations in the *CTNS* gene have been reported. This gene codes for cystinosis, which transports the amino acid cysteine. Cysteine crystals accumulate in the cells of many different organs, especially the kidneys and eyes, and lead to tissue and organ damage by forming toxic crystals. According to recent studies, cystine accumulation causes increased apoptosis, accumulation of distorted mitochondria, and increased oxidative stress, resulting in kidney cell dysfunction. Three clinical forms defined for cystinosis include neonatal nephropathic cystinosis and late or juvenile nephropathic cystinosis, and ocular or adult cystinosis. Neonatal cystinosis is the most common form of the disease with the most severe phenotype. Due to the hereditary and autosomal recessive nature of the disease, the lack of cost-effective definitive treatment, the progressive and destructive nature of the disease, and the high prevalence of consanguineous marriage in Iran and especially in West Azerbaijan Province, it is necessary to examine all introns and exons of *CTNS* gene in the patients in this province. The present research aimed to determine the frequency of pathogenic mutation c.971-12GA in intron 11 of the *CTNS* gene in nephropathic cystinosis patients referred to Shahid Motahari Educational-Therapeutic Hospital.

Methods and Materials: This study was conducted on patients with nephropathic cystinosis referred to Shahid Motahari Hospital in West Azerbaijan Province. Overall, 13 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 was taken from the patients in a 500 microliter EDTA-containing tube. DNA was isolated by salting out method from the entire blood of the tested patients. Intron 11 of the *CTNS* gene was amplified with specific primers. Then, PCR products were directly sequenced by an ABI 3700XL Genetic Analyzer. An in silico study was carried out to evaluate the variants. ACMG Guidelines (2015) were applied for the interpretation of the clinical significance of variants detected: pathogenic, possibly pathogenic, variant of uncertain significance, perhaps benign, and benign.

Results: This study investigated 13 patients with nephropathic cystinosis from 13 families to determine the frequency of pathogenic c.971-12ga mutation in intron 11 (rs375952052) in the *CTNS* gene.

Conclusion and Discussion: Our investigation failed to find the pathogenic mutation c.971-12GA in intron 11 of the *CTNS* gene in nephropathic cystinosis patients referred to Shahid Motahari Hospital.

Keywords: Genes, Cystinosis, Mutation

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