Turkish Journal of Pediatric Disease

Letter to the Editor

Comment on "Clinical and laboratory characteristics of nephropathic cystinosis in a resource-limited region"

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Received : 10.06.2026, Accepted : 30.06.2026 DOI: 10.12956/TJPD.2026.1384

Introduction

In the latest issue of the Turkish Journal of Pediatric Disease, Köksoy et al. (1) found that growth faltering and progression to renal failure were notable challenges for Turkish patients with cystinosis. We herein address a worthy notice. The CTNS gene mutations encoding a protein named cystinosine, which is a lysosomal cystine transporter, are the primary cause of cystinosis and there are approximately over 140 distinct CTNS mutations. The clinical phenotypes of cystinosis are controlled by the functional effects of the various CTNS mutations (2). Accordingly, sequencing the CTNS gene coding exons has been developed in numerous populations for the early cystinosis identification and characterization of the disease spectrum employing molecular diagnostic tests (3-4). Studies on the genetic landscape of cystinosis in Türkiye are scarce in the literature and they primarily involved small size series in localized settings (5-7). The available study that targeted CTNS mutations at national level based on a Turkish pediatric cystinosis registry was released by Topaloglu et al in 2017 (8). The 57-kb deletion, which is most common among Caucasian cystinosis patients, was not identified in any of the patients. However, seven novel mutations were detected. The most common CTNS mutations were c.681G.A (p.Glu227Glu), c.1015G.A (p.Gly339Arg), and c.18_21 del (p.Thr7Phefs*7) identified in 31%, 22%, and 14% of the patients respectively. Compared to other mutations, the above-mentioned three mutations were importantly correlated with the disease's earlier onset age and patients with severe CTNS mutations tended to develop worse renal outcomes (8). Notably, the identified genetic CTNS variants in Köksoy et al's study (1), namely c.681G>A (p.Glu227Glu), c.18_21delGACT (p.Thr7Phefs*7), and c.1015G>A (p.Gly339Arg), contribute to those previously addressed by Topaloglu et al (8). Updating national data on the CTNS mutational spectrum among cystinosis patients and genetic counseling where the families in Türkiye tend to be particularly large and display a high consanguinity rate (18.5%, with first cousin marriages accounting for 57.8%) (9) are deemed critical. These interventions together that recommended by by Köksoy et al (1), namely the initiation of cysteamine therapy, supporting access to the specialized healthcare facilities, and regular monitoring, are solicited to deter further breeding of cystinosis cases in Türkiye on one hand and improve patients' outcomes and quality of life on the other.

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