Al-Kindy College Medical Journal (KCMJ)

Letter to Editor

Comment on: "Neuropathic Cystinosis: A Rare Case Report"

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Article history:
Received 8 September 2025
Accepted 4 February 2026
Available online 1 April 2026

DOI: 10.47723/7h1d9798

Keywords: Cystinosis; CTNS gene mutation; Neurophysiological studies

Editor,

In the previous issue of the journal, Muthana et al[1] described a rare case of cystinosis in an Iraqi child with an initial neurological presentation. To further sharpen the case report's focus and expand the discussion, we present two relevant points.

First, Muthana et al.[1] reported that the child presented with poor growth and delayed acquisition of age-dependent milestones. The clinical examination revealed nothing significant except generalized hypotonia. Muthana et al.[1] appropriately referred to the genetic study in the studied patient and identified a deletion of 9-kb CTNS gene. Despite that crucial genetic test, which is known to be the only confirmatory diagnosis of the autosomal recessive nephropathic cystinosis[2], we think that neurophysiological studies in the form of electromyography (EMG) and nerve conduction studies (NCS) have to be included in the diagnostic protocol in evaluating the studied patient with hypotonia. Indeed, various neuromuscular involvements have been reported in cystinosis patients, such as clinically non-symptomatic proximal myopathy with an irregular pattern, isolated non-symptomatic sensory nerve conduction alterations, and combined abnormal sensory and motor axonal neuropathic alterations correlated with overt clinical findings[3]. If EMG and NCS were to be carried out, the findings could further enlarge the scope of neurophysiological changes in cystinosis patients described in the literature.

Second, CTNS gene mutations, which encode the lysosomal membrane cystine transporter cystinosin, are the eventual etiology of cystinosis. Worldwide, over 140 distinct CTNS mutations have been illustrated. There is a correlation between the clinical phenotypes and the functional outcomes of the different CTNS mutations [4]. As a result, sequencing the coding exons of the CTNS gene has been provided in certain populations for the early detection of cystinosis using molecular diagnostic investigations [5,6]. Although Muthana et al.[1] did not define the exact CTNS mutation in the studied patient, we think that the case report should stir up the need for mapping CTNS mutations in cystinosis patients in a greatly consanguineous Iraqi population. This step has important diagnostic, therapeutic, and preventive implications.

Funding

This study did not receive any specific funding from public, commercial, or non-profit organizations

Conflict of Interest

The authors declare no conflicts of interest related to this work.

Data availability

No datasets were generated or analyzed during the preparation of this commentary

Author Contributions

AAO and MJH contributed to the concept and drafting of the manuscript; HAA and SAM contributed to critical revision and intellectual content. All authors approved the final version.

All authors meet the ICMJE criteria for authorship and agree to be accountable for all aspects of the work.

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