Reviewer’s report

Title: CTNS mRNA MOLECULAR ANALYSIS REVEALED A NOVEL MUTATION IN A CHILD WITH INFANTILE NEPHROPATHIC CYSTINOSIS: A CASE REPORT

Version: 0 Date: 10 Sep 2019

Reviewer: John A. Sayer

Reviewer's report:

This is an interesting case report describing a novel mutation (large del) in a patient with cystinosis.

More detail is required regarding the initial gene tests. What did the report say - did exon 3 and 4 fail PCR, was MPLA or other investigations suggested at this time by the genetics centre. The delay in recognition and molecular genetic diagnosis is very important. What lessons can be learnt to prevent further delays in the future with similar cases?

What is the ethnicity of the family, was there any effort to find similar families - ie is this deletion a founder mutation?

Some clinical photos / data would be welcome as a new figure

How many cases of cystinosis are genetically unsolved or have just a single heterozygous change - could this deletion (heterozygously) allow other cases to be solved. Has the mutation been uploading into LOVD or a similar genetic database?

Please confirm that you have included your review in the ‘Comments to Author’ box?
As a minimum standard, please include a few sentences that outline what you think are the authors’ hypothesis/objectives, their main results, and the conclusions drawn. Your report should constructively instruct authors on how they can strengthen their paper to the point where it may be acceptable for publication, or provide detailed reasons as to why the manuscript does not fulfill our criteria for consideration. Please supply appropriate evidence using examples from the manuscript to substantiate your comments. Please break your comments into two bulleted or numbered sections: major and minor comments.

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