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: 19 Sep 2019

Reviewer: Roser Torra

Reviewer's report:

This a nicely written case report highlighting the use of unidentified mutations in the CTNS gene. I have very few comments.

Abstract:

Please mention why do you perform RNA analysis instead of DNA. It is explained in the manuscript but it sounds very strange in the abstract not explaining this, as RNA sequencing is not the usual first approach.

Careful with the name of the gene: CNTS instead of CTNS

Manuscript:

Mention if NGS could have detected the mutation without need of RNA analysis. Also consider mentioning that for suspected genetic diseases with no clear clinical diagnosis NGS may be of great help.

"Taking into consideration that cystine crystals can be determined after 16-18 months of age, we initially performed Sanger sequencing of the CTNS gene using genomic DNA which did not show anomalies because only part of the required genetic data were obtained."

This sentence is very confusing:

What's the relation between having crystals at 16-18 months and using genomic DNA?

What part of genetic data was missing?

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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NA

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NA

Are you able to assess the statistics?

- Are the statistical test(s) used in this study appropriate and well described?

- Is the exact sample size (n) reported for each experimental group/condition (as a number, not a range)?

- Are the description of any error bars and probability values appropriate?

- Are all error bars defined in the corresponding figure legends?

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