

## ESPS Peer-review Report

**Name of Journal:** World Journal of Nephrology

**ESPS Manuscript NO:** 3020

**Title:** Cystinosis as a lysosomal storage disease with multiple mutant alleles: Phenotypic-Gentotypic correlations

**Reviewer code:** 00503316

**Science editor:** Wang, Jin-Lei

**Date sent for review:** 2013-04-04 17:24

**Date reviewed:** 2013-04-15 22:40

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
<input type="checkbox"/> Grade A (Excellent)	<input type="checkbox"/> Grade A: Priority Publishing	Google Search:	<input type="checkbox"/> Accept
<input type="checkbox"/> Grade B (Very good)	<input checked="" type="checkbox"/> Grade B: minor language polishing	<input type="checkbox"/> Existed	<input type="checkbox"/> High priority for publication
<input checked="" type="checkbox"/> Grade C (Good)	<input type="checkbox"/> Grade C: a great deal of language polishing	<input type="checkbox"/> No records	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade D (Fair)		BPG Search:	<input type="checkbox"/> Minor revision
<input type="checkbox"/> Grade E (Poor)	<input type="checkbox"/> Grade D: rejected	<input type="checkbox"/> Existed	<input type="checkbox"/> Major revision
		<input type="checkbox"/> No records	

## COMMENTS TO AUTHORS

Review of WJN/2013 Editorial 1. The authors state: "hypothyroidism due to extensive cystine deposition had been reported and represent the cause of growth retardation of cystinosis patients". Growth failure starts long before hypothyroidism is evident and has multiple causes including chronic metabolic acidosis, electrolyte loss including (but not limited to) phosphatase loss causing rickets. 2. Proximal RTA non-related to Fanconi: Non-related should be changed to unrelated. 3. Gene should be written in italics 4. The extensive discussion of other forms of proximal RTA is interesting but is out of place in this paper and should be removed. A separate review of proximal RTA may be appropriate. 5. "In most patients, glomerular filtration rate remains normal for up to two years and then progressively deteriorates towards end stage renal disease (ESRD) at the end of the first decade" The authors should indicate that this applies to untreated patients. Those diagnosed early and treated appropriately do well. 6. The authors should discuss the new, slow release preparations of cysteamine. 7. The authors mention antenatal diagnosis but should also discuss pre-implantation genetic diagnosis.

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**Name of Journal:** World Journal of Nephrology

**ESPS Manuscript NO:** 3020

**Title:** Cystinosis as a lysosomal storage disease with multiple mutant alleles: Phenotypic-Gentotypic correlations

**Reviewer code:** 00503025

**Science editor:** Wang, Jin-Lei

**Date sent for review:** 2013-04-04 17:24

**Date reviewed:** 2013-04-16 04:48

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
<input type="checkbox"/> Grade A (Excellent)	<input type="checkbox"/> Grade A: Priority Publishing	Google Search:	<input type="checkbox"/> Accept
<input type="checkbox"/> Grade B (Very good)	<input type="checkbox"/> Grade B: minor language polishing	<input type="checkbox"/> Existed	<input type="checkbox"/> High priority for publication
<input type="checkbox"/> Grade C (Good)	<input checked="" type="checkbox"/> Grade C: a great deal of language polishing	<input type="checkbox"/> No records	<input type="checkbox"/> Rejection
<input checked="" type="checkbox"/> Grade D (Fair)		BPG Search:	<input type="checkbox"/> Minor revision
<input type="checkbox"/> Grade E (Poor)	<input type="checkbox"/> Grade D: rejected	<input type="checkbox"/> Existed	<input type="checkbox"/> Major revision
		<input type="checkbox"/> No records	

## COMMENTS TO AUTHORS

This review aims to highlight some data on a rare disease, with emphasis on its phenotype and genotype. I suggest to the author some modification to improve the article and to facilitate our reading: 1. The text needs to be proof read and carefully searched for misspellings 2. The author should better organize the information and consequently the structure of the paper. As suggestion, it will be better if the author could first describe the epidemiology of the disease, its genetic background, and its clinical presentation. Then, the author could point out specifically the renal presentation. After that, it will be easier if he could add the presentation of renal tubular acidosis but highlighting its connection with the disease, if not, it would sound abstract. 3. Improve the tables with clinical phenotype presentation, number of cases, sex and age, and treatment. 4. Improve the table 1 with reference. 5. Whenever, the author needs to summarize and to discuss the info presented in the literature, the controversies etc to give us a reasonable guideline of this disease.

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**Name of Journal:** World Journal of Nephrology

**ESPS Manuscript NO:** 3020

**Title:** Cystinosis as a lysosomal storage disease with multiple mutant alleles: Phenotypic-Genotypic correlations

**Reviewer code:** 00503428

**Science editor:** Wang, Jin-Lei

**Date sent for review:** 2013-04-04 17:24

**Date reviewed:** 2013-04-17 09:13

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
<input type="checkbox"/> Grade A (Excellent)	<input type="checkbox"/> Grade A: Priority Publishing	Google Search:	<input type="checkbox"/> Accept
<input type="checkbox"/> Grade B (Very good)	<input checked="" type="checkbox"/> Grade B: minor language polishing	<input type="checkbox"/> Existed	<input type="checkbox"/> High priority for publication
<input checked="" type="checkbox"/> Grade C (Good)	<input type="checkbox"/> Grade C: a great deal of language polishing	<input type="checkbox"/> No records	<input type="checkbox"/> Rejection
<input type="checkbox"/> Grade D (Fair)		BPG Search:	<input type="checkbox"/> Minor revision
<input type="checkbox"/> Grade E (Poor)	<input type="checkbox"/> Grade D: rejected	<input type="checkbox"/> Existed	<input type="checkbox"/> Major revision
		<input type="checkbox"/> No records	

## COMMENTS TO AUTHORS

I have reviewed the above numbered manuscript Cystinosis as a lysosomal storage disease with multiple mutant alleles: phenotypic-genotypic correlations by Mohammad Al-Haggar. The content is good. It is an uncommon disorder in the general practice of nephrology. However, the topic is important for a wide variety readers. Therefore it is acceptable but not in its present form. The article is somewhat repetitive hence, condensing the content, the article may prove to be meaningful. Therefore I recommend the following to improve its presentation; 1. Condense the inherited forms 2. Shorten the diagnosis of cystinosis in particular, with reference to the genetics. List the genetic mutations instead of the descriptive forms. 3. Reduce the references to relevant ones and no more than 25 to 30. I shall be pleased to review the revision. Kindly let me know your decision. Best regards.

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**Reviewer code:** 00503020

**Science editor:** Wang, Jin-Lei

**Date sent for review:** 2013-04-04 17:24

**Date reviewed:** 2013-04-23 12:10

CLASSIFICATION	LANGUAGE EVALUATION	RECOMMENDATION	CONCLUSION
<input type="checkbox"/> Grade A (Excellent)	<input type="checkbox"/> Grade A: Priority Publishing	Google Search:	<input type="checkbox"/> Accept
<input checked="" type="checkbox"/> Grade B (Very good)	<input checked="" type="checkbox"/> Grade B: minor language polishing	<input type="checkbox"/> Existed	<input type="checkbox"/> High priority for publication
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## COMMENTS TO AUTHORS

The manuscript is comprehensive but has some spelling errors and at times the English grammar could be improved.