Reviewer's report

Title: Phenotypic and Genetic Characterization of a Cohort of Pediatric Wilson Disease Patients

Version: 1 Date: 7 February 2011

Reviewer: George J Brewer

Reviewer's report:

1. (Major compulsory)
Third paragraph of Methods: Using 2 of the 6 criteria for diagnosis is unacceptable, and I suspect the authors didn't actually do that. For example a heterozygote could have a positive family history and 20% of them have a low ceruloplasmin. Please correct to your actual criteria.

Third par of Results: It says 69.2% positive KF rings, while the abstract says 97.6%. Please correct.

2. (Major essential)
5th par of Background: Zinc sulfate is mentioned as a treatment. Zinc acetate is used much more commonly around the world and is the FDA approved salt in the US as Galzin and is also approved in Europe as Wilzin. This should be pointed out.

Third par of Methods: A stain for copper is not very reliable for Wilson's diagnosis. Copper assay is the reliable method. The authors probably didn't have assay available, but they should clarify this difference.

3. (Discretionary)
2nd par of Treatment: A single daily dose of zinc is ineffective for copper control yet the authors used such a dose. Please explain why.

The authors referencing is sometimes inexplicable. For example ref 14 in 2nd par of Treatment Outcome doesn't seem to relate to the text. Similarly ref 7 in 5th par of Background is not a good ref for the topic. Much better would be Number XV of that series (Brewer et al J Lab Clin Med 132:264-278, 1998).

General Comment: The paper is a good description of Wilson's in Egypt, but is longer than it need be. The Discussion is long and duplicates things already discussed in Results. Also the Discussion lists many contrasts of Egyptian patients with those elsewhere and many are not significant and should probably be removed.

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable
Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I declare that I have no competing interests.