Reviewer's report

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

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Reviewer: Anna Czlonkowska

Reviewer's report:

The paper is interesting for everybody who is working with WD patients. Authors describe 77 patients with childhood onset of WD.

Results do not bring however much to our knowledge of the disease. Description of Egyptian cohort has a local value. The paper is generally poorly written so makes it difficult to follow.

Interesting is a high rate of consanguinity but authors did not give the definition of consanguinity.

Why authors are stating that disease is under diagnosed in Egypt. There is no information how many should be. They collected in 17 years 77 pediatric cases. The diagnosis is done only one year after symptoms onset which is not bad.

There are 4 different modes of therapy, so difficult to follow. Why they used D-p and zinc in those same persons. In that case measuring of copper in urine does not give information about effectiveness of treatment. How they could use urine copper as indicator of good treatment if at start they had data about copper level only from 25 cases.

I cannot understand many statements as “Of the 22 asymptomatic patients who were followed up, 19 had either a stable or an improving course”… What has improved in asymptomatic cases?

I do not know what to advice authors to do to make a paper easier. One way is to concentrate on smaller problems as genetics, biopsy. Interesting is one year old baby, how it was diagnosed?.

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

Quality of written English: Not suitable for publication unless extensively edited

Statistical review: Yes, but I do not feel adequately qualified to assess the statistics.

Declaration of competing interests:

'I declare that I have no competing interests'