Reviewer’s report

Title: A novel CISD2 intragenic deletion, optic neuropathy and platelet aggregation defect in Wolfram syndrome type 2

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Reviewer: Bruno Dallapiccola

Reviewer’s report:

This single case report describes a new patient homozygous for a mutation in CISD2 gene associated with Wolfram syndrome type 2 (WFS2).

At least 16 subjects, from four consanguineous Jordan families, have been clinically and molecularly characterized. Thus, the present study provides confirmatory results on the relationship between homozygous CISD2 mutations and WFS2.

The authors raise some concerns on the possibility that WFS2 is not a WFS subtype. This conclusion is based on the presence, in the reported patient, of optic neuropathy (ON), rather than optic atrophy (OA), which is considered and obligatory feature of WFS. They also suggest that OA in the Jordanian cases could be in fact ON. However, this statement has no support in the clinical study published in 2000, nor in the clinical supplement to the molecular study in 2007. Rather, intestinal ulcers could represent a distinguishing feature in the two WFS types. Since this manuscript does not provide significant novelties, I suggest the authors to discuss more in deep the clinical features possibly differentiating the heterogeneous spectrum of WFS and eventually summarize them in a Table.

In addition, I also recommend the authors to provide some data concerning the geographical origin of the patient’s parents.

Level of interest: An article of limited interest

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