Reviewer's report

Title: The Clinical And Molecular Spectrum Of Galactosemia In Patients From The Cape Town Region Of South Africa

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Reviewer: Prof Juergen Reichardt

Level of interest: A paper whose findings are important to those with closely related research interests

Advice on publication: Accept after discretionary revisions

The manuscript by Henderson and colleagues investigate the frequency and molecular basis of galactosemia in South Africa. This population is to-date only poorly characterized. The methods are well described and the authors' data support their conclusions.

Henderson et al. should address the following issues:

1. M&M: The authors must provide references for their biochemical assay methodologies.
2. M&M: References for the Q1888R and S135L mutations and the accompanying frequency statements must be included.
3. M&M: Why did Henderson et al. use two different assays for genotyping the S135L variant. The reproducibility of the two methodologies must be presented, compared and discussed (if there were discrepancies).
4. Discussion: Henderson and colleagues surely do not mean that the "carrier frequency" of galactosemia has been reported as "1/30,000 to 1/50,000" by others. They presumably mean incidence figures and must correct this sentence appropriately.
5. Discussion: Beutler et al. (referenced by the authors) reported a frequency of 0.1% for the S135L variant in African-Americans. Others (e.g. Ng et al.) have also reported on this mutation in the US. These data should be discussed within the context of the authors' finding of a 1/60 frequency for galactosemia carriers amongst Africans in South Africa.
6. Table 1 (and text): Reporting % figures with the small number of patients investigated seems like a bit of a stretch. Henderson et al. should use more precise language and data.
7. Fig. 1 does not seem to add any new information to the galactosemia literature.

Competing interests:

None declared.