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 Derek Applegarth

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

Advice on publication: Accept after discretionary revisions

Comments

Compulsory revisions

Page 4, line 2 down
Insert "to" between "home" and "3.15".

Page 9, line 5 up
Delete the words "at least" and add the word "approximately".

Page 11, line 1
Delete the word "frequency" and add the word "incidence".

Page 11, line 3 down
Delete the word "frequency" and add the word "incidence".

Page 11, line 5 down
Delete the word "frequency" and add the word "incidence".
In the second last line of the same paragraph delete the word "to" and add the word "in".

Discretionary revisions

I suggest that the authors add a sentence in their conclusions section which would state that the apparent carrier frequency obtained by investigating 725 healthy black newborns of approximately 1 in 60 for the Cape Town metropolitan area is very close to the approximate carrier frequency reported by Manga, et al., (reference 8) for a more northern area of S. Africa of approximately 1 in 75.
The authors describe the presence of conjugated hyperbilirubinemia in patients who had a mean age of diagnosis of about 5.1 months. I have frequently seen unconjugated hyperbilirubinemia in children who are diagnosed in the neonatal period. Therefore, on page 8, paragraph 2, line 6 down it would probably be a good idea to consider adding the words "or mixed" after the word "conjugated".

On Page 8, paragraph 2, last line it would probably be better to delete the word "is" and add the words "may be" because the clinical manifestations of the S135L mutation are incompletely described in the medical literature. Some individuals who are homozygous for this mutation appear to have a considerable ability to metabolize galactose so it is hard to know whether or not there are some homozygotes in the South African population who may have such a mild clinical presentation that they may have been missed. Equally, there may have been severely effected patients who may have died earlier and, therefore, been missed.

**Competing interests:**

None declared.