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: Dr LJ Elsas

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

Advice on publication: Accept after discretionary revisions

This is a descriptive review of a galactosemic population in Africa and supports previous observations that the S135L mutation is found in Africa and the Qi88R mutation in GALT is found predominantly in caucasians of Northern European descent. It would be stronger if there were more data presented on the clinical variation among the S135L/S135L, Q188R/Q188R, and heterozygous patients. Were previous observations that the S135L mutation has less clinical impact than the Q188R upheld? What were the relative neonatal and longer term outcomes among these three genotypes? Were there other mutations identified?

Competing interests:

None declared.