Reviewer’s report

Title: Screening for inborn errors of metabolism in high risk children: a three-year study in Zhejiang Province, China

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Reviewer: Rita Christopher

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Tandem mass spectrometry is an effective tool for screening multiple inborn errors of metabolism in a single analysis. Huang and coworkers have used this technique to screen 11,060 high-risk children from the Zhejiang Province, China, and detected 62 cases during a 3-year period.

Major Compulsory Revisions

The authors have mentioned that the MS-MS analysis has been performed according to the protocol provided by the manufacturers. The name of the manufacturer and some details of the assay procedure could be provided.

Blood acylcarnitine and amino acid concentrations are known to alter with age. The authors have established the cutoff values by a study on full-term newborns. The median age of the patients in their study is 28.8 months. Since age-specific cut-off values have not been used, this could have resulted in errors during interpretation. The authors need to address this issue suitably.

Adequate data to support the method by which the positive cases have been confirmed should be provided.

The authors have reported 3 cases with ornithine transcarbamylase (OTC) deficiency, a proximal urea cycle defect. One of the notable deficiencies in the use of tandem mass spectrometry is the inability to screen proximal urea cycle disorders, including OTC defects. The criteria used for identifying OTC cases and methods used for confirming the diagnosis should be mentioned.

The limitations of the study should be stated clearly.

The text of the manuscript has grammatical and spelling mistakes and should be extensively edited.

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

Quality of written English: Needs some language corrections before being published

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