Reviewer's report

Title: Imparting carrier status results detected by universal newborn screening for sickle cell and cystic fibrosis in England: a qualitative study of current practice and policy challenges

Version: 2 Date: 10 June 2007

Reviewer: Bridget Wilcken

Reviewer's report:

General
The authors have carried out a limited qualitative study to discover current practice in England of how carrier testing results obtained as a "by-product" of newborn screening are conveyed to parents. This is useful and timely. The manuscript is interesting, but is a little diffuse, and might be improved by tightening the writing in some places. Some areas noted below may also improve the paper.

Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)
1) Background paragraph 2. The description of the screening protocol for CF is not at all clear. It should be made clear that it relates to screening protocols for CF in England, as there are many different protocols elsewhere. Additionally, for clarity, the authors need to include the elements that a screening test for immunoreactive trypsin identifies those babies at high risk for CF, and these samples are further tested for a small panel of CF mutations. Those with 2 disease causing ones will have CF, but those with only one identified mutation could have CF, having another mutation, or could merely be carriers, and would need a repeat newborn screening test at 21-28 days, when it is more discriminatory.

Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)
1) p5 A role for non-specialists. Here it was easy for me to miss reading the footnote, and then to wonder what was meant by specialists etc. The footnote should be incorporated in the body of the text.

Discretionary Revisions (which the author can choose to ignore)
1) The quotes included in the results section are helpful. However, perhaps they need not be quite so verbatim. eg: p5 "We had a case...where we found 60 children hadn't been given results....because people didn't quite know what to do..." might be an improvement. Similarly, using "erm" was rather unnecessary and interruptive.
2) As mentioned above, the writing could be tightened throughout, without losing any important aspects.

What next?: Accept after minor essential revisions

Level of interest: An article of importance in its field

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