Reviewer’s report

Title: Identification of novel isoforms in the assessment of SNF2L as an XLMR (X-linked mental retardation) candidate gene in 12 families linked to Xq25-26.

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Reviewer: Gajja S. Salomons

Reviewer’s report:

The study describes:
1) the genomic DNA sequence analysis of the SNF2L gene in 12 patients linked to Xq25-26, but no mutations could be detected
2) the expression pattern by RT-PCR of 2 isoforms of SNF2L that have been published in 1992 by a different group
3) the presence and absence of NLS in the SNF2L gene. However, it is unclear which splice variant does or does not contain this deletion.

The problem with this paper is that it does not have a focus, and consists of 3 main topics. Each of these topics appear not to be investigated in depth and the direct link between the subjects is not obvious.

ad 1) The hypothesis that SNF2L is a candidate XLMR gene appears to be raised by the authors in previous publications and warrants follow-up studies. The fact that no mutation was identified in only 12 patients does not rule out this hypothesis. Actually the study would gain much more power if a large panel of presumed XLMR patients would be investigated (e.g. more than 300 patients from the EUROMRX consortium/ Dr Hans van Bokhoven and/or Greenwood Genetic Center/ Dr C Schwartz, as these groups already provided the 12 linked XLMR families)

ad 3) How is the NLS characterized? See Results and Discussion section, 2nd paragraph “we noticed that”-. Here it could be stated in more detail that the human (which?) and mouse SNF2L genes were analyzed by blast tool. As mentioned above the presence and absence of NLS in the SNF2L gene has been investigated. However, it is unclear which splice variant does or does not contain this deletion. These findings are likely novel, but this is difficult to digest by the reader.

What next?: Reject because too small an advance to publish

Level of interest: An article of limited interest

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