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.

**Version:** 1  **Date:** 23 November 2007

**Reviewer:** Takahito Wada

**Reviewer's report:**

**General**

This is a paper about a candidate gene, SNFL2, encoding an ATP-dependent chromatin remodeling protein for X-linked mental retardation. Mutation of genes involving in chromatin remodeling or epigenetics is one of the most important causes for mental retardation. So this paper seems fascinate from the point that the new chromatin remodeling gene, SNFL2, could be a candidate gene for mental retardation.

Although the author did not find any mutations in MR patients, this paper may shed light on chromatin remodeling proteins and mental retardation, and should be appreciated. This manuscript paper is suitable for publication in the BMC Medical Genetics.

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**Major Compulsory Revisions** (that the author must respond to before a decision on publication can be reached)

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**Minor Essential Revisions** (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

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**Discretionary Revisions** (which the author can choose to ignore)

1. Are there any candidate genes for mental retardation, which map to Xq25-26? If so, have you analyzed their genes for your patients?
2. Did you do X inactivation study for an obligatory carrier female with normal IQ?
3. As you mentioned in “Conclusions”, you cannot exclude intronic mutations, affecting alternative splicing. Can you confirm this by doing RT-PCR using cell lines of patients?
4. Is it possible to analyze protein level for your patients’ diagnosis?
5. As you see in ATR-X syndrome, a mutation of genes coding chromatin remodeling protein can show various clinical phenotype. How about SNF2L?
What next?: Accept after minor essential revisions

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

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.