Reviewer’s report

Title: Genotype-phenotype correlation of proximal tubular function in Imerslund-Gräsbeck Syndrome

Version: 2 Date: 20 November 2012

Reviewer: Sandya Liyanarachchi

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Authors investigate 9 patients with Imerslund-Gräsbeck Syndrome (IGS), a rare genetic disorder. They identified new mutations and previously described mutations in these patients and performed functional characterization of the mutations. Urinary protein excretions of the patients were evaluated. Provide evidence for a correlation between mutation type and the presence of low-molecular-weight proteinuria observed in most patients.

Compulsory revisions:
1. The claim of “Genotype-phenotype correlation…..” in the title seems to be more general and a stronger statement than the observed results. Better to have a title that represents the actual strength of the observed results.
2. Better to add more information to improve Figure 2 and its legend. For example, indicate in the figure or legend what is represented by lower case letters, what is the arrow for, indicate the stop codon after the frame shift, ……..

Minor revisions.
1. Indicate the reference for the mutation FM1 on page 5.
2. In Figure 3, figure label overlaps with “ORF CONTROL”.
3. Table 2, the last row and last column “c” should be “C"
4. On page 26, Table 2 legend, there is an extra “)“ in the sentence below. ………..cubilin and megalin ligands, see Table 3).

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.