Reviewer’s report

Title: Ser80Ile mutation and a concurrent Pro25Leu variant of the VHL gene in an extended Hungarian von Hippel-Lindau family

Version: 2 Date: 29 February 2008

Reviewer: Rob B van der Luijt

Reviewer’s report:

- Major Compulsory Revisions
The authors have adequately addressed the original comments 1, 2 and 3. Comment 4 has not yet been fully resolved, requires a Minor Revision (original comment 4: The authors do not use the standard HGVS nomenclature for the description of sequence variants, while it is generally recommended to do so. At least, the authors should provide a full description of the VHL changes identified in their patients, i.e. including the nucleotide changes as well as the predicted amino acid changes).

Although in the revised version of the manuscript, the authors have provided the nucleotide changes of both VHL sequence variants by including the base triplets corresponding to the respective codons, the nucleotide numbers have not been included. To avoid confusion, the nomenclature should be modified as follows (see Table).

Nomenclature by authors in revised MS Nomenclature based on HGVS recommendations
AGT80AAT (Ser80Ile) c.239G>A, p.Ser80Ile
CCT25CTT (Pro25Leu) c.74C>T, p.Pro25Leu

- Minor Essential Revisions: all previous comments were adequately addressed.
For the revised version, please note the following minor essential revision
Page 8, line 3: â##tropicalsâ## should be changed into â##tropicalisâ##

- All Discretionary Revisions were adequately addressed

Based on your assessment of the validity of the manuscript, what do you advise should be the next step?

- Accept after minor essential revisions

For (1) Level of interest, (2) Quality of written English, (3) Statistical review, (4) Declaration of competing interests please see my original review dated December 11th, 2007.