Reviewer's report

Title: Clinical and genetic analyses of three Korean families with hereditary hemorrhagic telangiectasia

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Reviewer: gaetan lesca

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Kim et al., report on the clinical and genetic data of three Korean families with HHT. This is the second description of HHT families with molecular confirmation in Korea. They found three mutations, 2 in ENG, including a novel one and 1 in ACVRL1. For the c.1-127C>T mutation, the authors performed very interesting experiments showing that it can prevent translation of ENG. The discussion is well written.

Minor essential revisions:

In the “Background” section:
- I would add other references concerning the mutation rate of ENG and ACVRL1 in HHT (For exemple, Lesca et al., Genet Med 2006).
- The date of the last update of the HHT database should be mentioned.
- Recent data by the team of S Bailly strongly suggesting the role of BMP9 as a (the) major ligand of ALK1 and Endoglin should also be mentioned.
- I think “incomplete penetration” should be replaced by “incomplete penetrance”

In the “methods” section, I would replace “affected immediate family member” by “affected first degree family member”.

In the “results” section, the description of the families should be improved:
- The number of patients having telangiectases seems surprisingly low (1 patient!) , in contradiction with the “Background” section. Were all the family members carefully examined by a trained clinician?
- Some patients had seizures and embolic cerebral infarction or cerebral abcess. Were seizures the revealing symptoms of infaction and abcess?
- Were the PAVMs found in patients due to clinical symptoms (embolism, infarction, dyspnea,…) or were some of them found by systematic screening?
- Which methods (MRI, X ray, angiography…) were used for clinical evaluation of AVMs?
- Were patients screened for Hepatic AVMs?
- For family 1, it is mentioned that “two children of the affected proband’s sister (I-3) were diagnosed free from HHT”. Does it mean that they had no symptoms or that they did not bear the family mutation?
Discretionary revisions

The second sentence of the conclusion is not very informative.

**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