Reviewer's report

Title: Compound heterozygosity of predicted loss-of-function DES variants in a family with recessive desminopathy

Version: 1 Date: 17 March 2013

Reviewer: denis duboc

Reviewer's report:

The method and the results are of interest. (The clinical report is clearly presented)

Some reference concerning the autosomal recessive inheritance are lacking and could improve the quality of the discussion which is the major interest of the case report.

A recent review published in Jan 2013 (or other) on mechanism and pathology of desminopathies could be helpful

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'