Reviewer's report

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

Version: 1 Date: 22 March 2013

Reviewer: Anna Kaminska

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Minor Essential Revisions

As for the genetic part the authors could more rigorously substantiate their clam about the causative influence of both mutations. For instance they could provide an analysis of DES transcripts, to show stable, truncated DES mRNA, together with full-length mRNA. They could also show at the protein level the presence of both normal and truncated DES polypeptide forms.

Level of interest: An article of importance in its field

Quality of written English: Not suitable for publication unless extensively edited

Statistical review: No, the manuscript does not need to be seen by a statistician.