Reviewer’s report

Title: LMNA mutations in Polish patients with dilated cardiomyopathy: prevalence, clinical characteristics, and in vitro studies

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Reviewer: Joakim Klar

Reviewer’s report:

In this manuscript the authors describe the finding of LMNA mutations in patients with dilated cardiomyopathy, including a few novel ones.

Minor Essential Revisions

1. Mention the GenBank reference sequence and version number for the LMNA transcript studied.

2. The pathogenicity p.Val256Gly mutation was proposed to be proven in part by the conservation of the p.Val256 amino acid. However, prediction programs (e.g. PolyPhen2) suggest that this variant would be benign. Instead it is nicely shown that mutant protein forms lamin aggregates in a cell system, which would in this case prove the pathogenicity. What is known about lamin aggregate formations for the other mutations (non-sense versus missense).

3. Could the authors comment on the severity of the phenotype for different mutations (non-sense, missense, and protein domains).

4. Write systematic names for both DNA and protein variations at the first mentioning in the methods. This will make it easier to follow how the mutations where screened for at the DNA level. Also, this could also be included in the Figure 1 text, as this describes the variants at the DNA level.

5. Describe what the arrow indicate in Figure 4B (focal breakage) and perhaps add arrows to Figure 4A and C to draw the readers’ attention to the described changes of the nuclei.

6. The level of magnification used should be added to the text of Figure 5.

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