Reviewer’s report

Title: Whole exome sequencing identifies a novel EMD mutation in a Chinese family with dilated cardiomyopathy

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Reviewer: Marcella Neri

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The Authors describe a huge Chinese family with DCM and an X linked pattern of inheritance; they used a combined approach of linkage analysis and whole exome sequencing to identify the disease gene.

Minor Essential Revisions

1 in the background they reported 97 EMD mutations known to date (HGMD) but in the LOVD gene specific database the DNA variants are 223. Among these variants there are several deletions, in particular involving exon 1 the “c. 3_31 del” is recurrent and associated to a constant cardiac involvement. It is worth to discuss these informations and to speculate a possible role of 5' protein region in the prevalent cardiac phenotype.

2 the muscular phenotype of one of the patients, evaluated after the identification of EMD mutation, is only clinically described. A CK evaluation or electrophysiological (EMG) evaluation or CT scan of muscles would be necessary to define the skeletal disorder as “mild”.

3 in the Methods, exome sequencing, is not specified how was constructed the library: which kit was used? could be responsible for the the sequencing failure detected?

4 in the results of WES is written “no significant variants in the exons of all reported non syndromic DCM causing genes” and the reference is Hershberger 2011. I would add a table with the list of the genes screened and compared it with the genes reported in the http://www.musclegenetable.fr (updated to 2013)

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