Reviewer's report

Title: A study in Polish patients with cardiomyopathy emphasizes pathogenicity of phospholamban (PLN) mutations at amino acid position 9 and low penetrance of heterozygous null PLN mutations

Version: 1 Date: 13 November 2014

Reviewer: Madhu Khullar

Reviewer's report:

Major compulsory revisions: None

Minor Essential Revisions: Authors have discussed that c.116A>C:p.Leu39Ter (rs111033560), a null variant of PLN leads to truncated protein and is pathogenic, and extrapolate these findings to c.9_10insA:(p.Val4Serfs*16) null variant identified by them in their cohort, the supporting laboratory data for this is not provided. In fact this variant does not appear to be associated with cardiomyopathy, hence a general statement “our results and the data reported in the literature indicate that in humans 213 heterozygous null PLN mutations either have low penetrance or are not pathogenic” may not be correct and should be modified accordingly.

Discretionary revisions: None

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'