Reviewer's report

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

Version: 1 Date: 4 February 2013

Reviewer: Nicola Carboni

Reviewer's report:

minor essential revisions:

Background, page 3. The list of diseases associated to LMNA gene alterations should include more diseases: i.e. MADA, HHS (heart hand syndrome of Slovenian type et cetera) and also should be cited w references the overlapping syndromes due to LMNA gene mutations. In the aforesaid list, it is reported DCM; this is clearly wrong since the correct name is dilated cardiomyopathy with conduction defects.

page 4, line 10: 215 Caucasian subjects with no history of cardiovascular diseases....; "Does the population include tis w muscular dystrophy or other diseases possibly linked to LMNA gene mutations?"

Same page: Mutations screening: where flanking intronic regions explored?

Page 5, Expression analysis: please add a citation of manufacturer protocol (line 13).

Clinical evaluation of the mutation carriers is really redundant. The description of index cases clinical conditions should be better summarised in a table reporting gene mutations and clinical manifestations.

Discussion: page 9, last line: Synus node dysfunction at the onset of the disease is not typical....; "please support this observations w data already published or delete it. Actually, there are reports describing such a modality of diseases presentation.

page 11, first line: His EDMD symptoms did not appear .....; Is this phenotype in keeping w EDMD? Are you sure? Any detection of elbows, TA contractures? rigid spine? More likely LGMD1B.

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 I have no competing interests below