Reviewer's report

Title: Cell stress molecules in the skeletal muscle of GNE myopathy

Version: 2 Date: 15 November 2012

Reviewer: Boel De Paepe

Reviewer's report:

It was with great interest that I read the manuscript by Fischer et al. entitled “cell stress molecules in the skeletal muscle of GNE myopathy”. The text is well written and addresses an interesting and timely topic. Some points could allow to improve the paper further:

Major compulsory revisions

1. On page 7, it is mentioned that MHC-I expression remained at a slightly lower level. Please be more specific and less vague.

2. The results of expression studies that are summed up on page 7 state ‘higher expression’, yet no statistical significance was reached. Please state this important nuance specifically in the text.

3. Figure 1 does not show mean levels, which makes it difficult to interpret possible differences. Please introduce marks for mean levels.

4. The discussion begins with the statement that alphaB-crystallin and iNOS are the most prevalent cell stress markers in GNE myopathy. Please rewrite this sentence, as only a selection of stress factors was tested for this manuscript.

5. Why did the authors choose to test pro-inflammatory cytokines, although GNE myopathy is generally not associated with muscle inflammation? Also, many of these factors are present in control samples, which is an argument for a possible physiological role. Please comment.

6. In figure 3, iNOS staining appears generalized, yet the authors scored fibers positive and negative. Please elaborate on the scoring (what is regarded as positive?) in the methods section.

Discretionary revisions

1. Why was exact age of patients not provided in table 1?

Level of interest: An article of outstanding merit and interest in its field

Quality of written English: Acceptable

Statistical review: Yes, but I do not feel adequately qualified to assess the statistics.

Declaration of competing interests:
I declare that I have no competing interests