Reviewer’s report

Title: Exome sequencing identifies titin mutations causing hereditary myopathy with early respiratory failure (HMERF) in families of diverse ethnic origins

Version: 1 Date: 27 November 2012

Reviewer: Hans Goebel

Reviewer’s report:

This is a very comprehensive and pertinent paper concerning MFM. It is clearly written, well illustrated and superbly edited. Only few minor points, largely misprints, require attention:

1. Several gaps in brackets (..) require additional information or closing the gaps: pp. 10, 15, 38
2. p.11. lines 7 and 15 : "..year-old.."
3. p.12, line 2 from below : "..enzyme activities."
4. p.18,line11 : "..sternocleido..."
5. p.19, line 9:"..sarcolemma.."; line 4 from below : PAM (protein aggregate myopathies), because PA diseases are also neurodegenerative ones, such as Alzheimer etc.
6. p.28, ref.26 : "et al." not in bold, see refs.23 and 27
7. p.32, Table 1 : "shoulder adduction"
8. p.36, legend Fig. 3.: "Histological and histochemical..", because H&E and trichrome are stains; "fibrofatty"; "occupied"; Fig.- 4.: "..analysis of muscle tissue in patients.."
9. p.37, line 1 : _ "..images of muscle fibers in patients..."

Level of interest: An article of outstanding merit and interest 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 have no competing interests