Reviewer's report

Title: Two novel missense mutations in the myostatin gene identified in Japanese patients with Duchenne muscular dystrophy

Version: 3 Date: 12 February 2007

Reviewer: Jean-Claude Kaplan

Reviewer's report:

General

This revised version has adequately taken into account of the reviewers' remarks. It is interesting that a variation (2 nt del) was found in the 3'UTR upon additional sequencing. The relevance of this finding deserves further studies in another paper.

In its present state the paper is OK for publication under the provision that the authors modify their persisting wrong assessment (page 11 of the ms) that the heterozygous mother of the propositus of Schuelke's paper was asymptomatic. In fact in this paper one reads on the first page (page 2682) that she "was a former professional athlete" and on the following page that « she appeared muscular though not to the extent observed in her son » . In addition the family tree in Fig 1 of Schuelke's paper clearly shows that the heterozygous members are not asymptomatic, since they are reported as being « exceptionnally strong ».

On page 12 of the manuscript the text of lines 3 to 7 is not understandable, because ther is no verb in the sentence starting with « ConsideringŠ », as if some words were missing.

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

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Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

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Discretionary Revisions (which the author can choose to ignore)