Reviewer’s report

Title: The Facioscapulohumeral muscular dystrophy region on 4qter and the homologous locus on 10qter evolved independently under different evolutionary pressure.

Version: 2 Date: 23 October 2006

Reviewer: Yukiko Hayashi

Reviewer’s report:

General
The authors characterized chromosomal structure of 4q35 FSHD region and its highly homologous 10q26 region in a large number of FSHD patients and normal individuals. There are several important cases are included. Presence of BlnI-resistant qA-type fragments between 30-39 kb in normal individuals is important. Also, the presence of qA-type of 33 kb and 38 kb fragments observed in the FSHD patients with two 4q-linked short fragments and their non-affected family members. It is important to know the clinical difference between these two patients with two short fragments and the other patients (or family members) with one short fragment. These results suggest us a careful consideration to make a diagnosis for FSHD if the fragment size are between 30-39 kb.

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

The presence of a healthy individual with deletion from the probe p13E-11 and whole D4Z4 repeats, and a healthy subject (N5) with a variant 4q allele containing less than 10 repeats are also quite important. Additional discussion is helpful to consider the pathomechanisms of FSHD.

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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)

In the Background, the authors wrote ?The reduction in the number of 3.3 kb D4Z4 repeat to an array of 1-11 units at the 4q35 locus is tightly?.?. The number should be changed to 1-10 to avoid confusion, since in the RESULT section, they describe ??the presence of a number of D4Z4 above the threshold number of 10 do not leads?.?.

The initial half of the enzyme name should be described in italic.
In the first paragraph of RESULTS, nullisomy should be added in the names of the allelic pattern.

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Discretionary Revisions (which the author can choose to ignore)

What next?: Accept after minor essential revisions

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: No