Reviewer's report

Title: Facioscapulohumeral Muscular Dystrophy and Charcot-Marie-Tooth Neuropathy 1A - Evidence for "double-trouble" Overlapping Syndromes

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Reviewer: Rossella Tupler

Reviewer's report:

This interesting article reports on a patient with genetically confirmed overlapping diagnoses of CMT1A and FSHD. The authors highlight the concept that in FSHD, even if a mutation the major disease locus has been found, further genetic testing might be warranted in cases with unusual clinical presentation having direct impact on medical care and genetic counseling.

This is especially true for FSHD where several unusual phenotypes and atypical morphological features have been previously described, also in association with pathogenic mutations in other genes. The patient had no facial weakness and negative family history for myopathies.

The article is well written and the case clearly described.

I have only few suggestions/questions for the Authors:

1. Page 7 “Case description Clinical data” The authors should describe the motor impairment of shoulder girdle in detail. For instance they should specify that the patient was unable to lift his arms above the horizontal level as reported in figure 1 legend.

2. Page 8 The Authors reported that HE staining of a muscle biopsy showed moderate myopathic changes indicated by numerous hypertrophic type I fibres. However, Figure 4 shows grouped fiber atrophy that can be indicative of neurogenic changes. Were also observed fiber type grouping? A picture of ATPase staining should be added in order to support the description of changes in different types of muscle fibers.

3. Page 8 “Genetic analysis” should be changed into “molecular analysis” since family members have not been investigated.

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 that I have no competing interests.