Reviewer's report

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

**Version:** 4  **Date:** 18 May 2013

**Reviewer:** Davide Pareyson

**Reviewer's report:**

This is an interesting description of a patient with a casual and unlucky co-occurrence of FSHD and CMT1A. The case report is well reported and discussed.

Major compulsory revision.

1) This is another example of a patient harbouring the FSHD-associated deletion of the D4Z4 repeat and showing an atypical phenotype due to a concurrent disease. Very recently the pathogenic relevance of the deletion have been seriously questioned (Scionti et al. Large-scale population analysis challenges the current criteria for the molecular diagnosis of fascioscapulohumeral muscular dystrophy. Am J Hum Genet 2012;90:628-35) and this case might provide some further material for discussion.

As this is a single sporadic case and the validity of the molecular criteria for FSHD diagnosis are under criticism, some caution is required.


Minor essential revision

3) Four major diagnostic criteria for diagnosis of FSHD are detailed on pages 4-5. However, criteria # 2 (facial weakness) and # 3 (autosomal dominant inheritance) are not satisfied by the patient.

4) It would be important to describe needle EMG findings (myopathic and/or neurogenic pattern?).

Discretionary revision

5) It is not clear which were the myopathic changes at muscle biopsy. Were there other changes beyond hypertrophic type I fibres? I cannot see the nuclei in figure...
4 (muscle biopsy).

**Level of interest:** An article whose findings are important to those with closely related research interests

**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