Author's response to reviews

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

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Dear Mr. Sands,

Enclosed please find our manuscript entitled: “Facioscapulohumeral Muscular Dystrophy and Charcot-Marie-Tooth Neuropathy 1A - Evidence for "double-trouble" Overlapping Syndromes” for consideration for publication as research article in BMC Medical Genetics.

We report on a patient with genetically confirmed overlapping diagnoses of hereditary motor and sensory neuropathy type 1A (CMT1A) due to a PMP22 gene duplication and facioscapulohumeral muscular dystrophy (FSHD) due to a partial deletion of the D4Z4 locus (19 kb). Both are rare diseases with a prevalence of approximately 1:5.000 (CMT1A) and 1:20.000 (FSHD), reflecting that a patient who suffers from both diseases can be found with a frequency of one in 100 million people.

This case adds to the increasing number of unique patients presenting with atypical and overlapping phenotypes in neuromuscular disorders. Even if a mutation in a disease gene has been identified, further genetic testing might be warranted in cases with unusual clinical presentation due to direct impact on patients’ benefit from adequate medical care and appropriate genetic counseling, especially in the light of arising causative therapies.

All coauthors have seen and agreed with the contents of the manuscript. The submission is not under review at any other journal. There are no competing interests to be declared.
We hope that this report is of interest to the readership of BMC Medical Genetics.

With best regards

Sincerely yours,

Maggie Walter