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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Author's response to reviews: see over
Dear Professor Neri, dear Editorial Board,

Enclosed please find our revised manuscript in which we addressed the reviewer’s comment concerning the calculation of the estimated frequency of the described coincidence of CMT1A and FSDH in the same patient.

**Reviewer’s comment**: I would calculate the likelihood of being affected by both FSDH and CMT in 1:50,000,000. Can you please check your calculation?

We changed the sentence on page 9, 1st paragraph, accordingly:

“Recent epidemiological data suggest that the frequency of CMT1A is around 1:7,500 [4] while the frequency of FSHD is estimated at 1:20,000 [7]. Thus, the chance of being affected by both disorders is about 1:150,000,000.”

In order to show recent epidemiological data on CMT, we added this reference:


With regard to these new data from Gess et al., we also adapted the first sentence in our background section on page 4:

“Hereditary motor and sensory neuropathy (HMSN), also called Charcot-Marie-Tooth (CMT) disease, is the most common inherited neuromuscular disorder with an estimated prevalence of 1:2,500 [1]. Roughly one third of all cases are caused by an autosomal dominant inherited 1.5 Megabase (Mb) tandem duplication encompassing the peripheral myelin protein 22 gene (PMP22) on chromosome 17p11.2-12 which encodes an important component of peripheral nervous system myelin [2-4].”

We hope that you agree with the improvement of our manuscript and thank you for your time and the acceptance for publication.

With best regards, sincerely yours, Maggie Walter