Severe axonal Charcot-Marie-Tooth disease with proximal weakness caused by de novo mutation in the MORC2 gene.

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Sir,

It was with great interest that we read the article on advanced access by Sevilla et al. (2015) regarding axonal Charcot-Marie-Tooth (CMT2) disease caused by mutations in the MORC2 gene. Through whole-exome sequencing in a Spanish four generation CMT2 family with autosomal dominant pattern of inheritance, the authors identified the mutation p.R190W in the MORC2 gene as the cause of the disease. It was the only variant detected by whole-exome sequencing that segregated with the disease in the family. Afterwards, they tested an additional 52 patients with axonal Charcot-Marie-Tooth and found mutations in the MORC2 gene in two additional families, one with different de novo mutation and the second with the same p.R190W mutation, also de novo.

Here, we would like to report another patient with the p.R190W mutation in the MORC2
gene and thus confirm the causality of this gene for the severe CMT2 with striking proximal weakness. Also, we would like to show that the p.R190W mutation is a hot spot and probably the most frequent mutation in this gene. Immediately after we became aware of this newly discovered gene, we detected this mutation by re-examining older whole-exome sequencing data in one of...


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