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Different nerve excitability measurements in a rare case of CMT1H

12/ Nerve and muscle excitability

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Objectives

To document electrophysiological abnormalities in a paucisymptomatic patient carrying a pathogenic heterozygous mutation c.1117C>T (p.Arg373Cys) of the FBLN5 gene.

Content

Methods:
Conventional nerve conduction studies, and motor axonal excitability evaluations of the median nerve at the wrist including threshold tracking (TT) and manual measurements (MM) of rheobase, strength-duration time constant (SDTC), iMAX parameters (for ulnar and fibular nerves also), refractory period and early-supernormal and late-subnormal periods were done. In addition, an ultrasound evaluation of the median nerve was performed.
Results:
Conventional nerve conduction studies revealed a homogenous sensory-motor demyelinating polyneuropathy (median nerve motor conduction velocity < 38 m/s), without significative sensory-motor axonal loss. The three parameters of the iMAX procedure (minimal threshold, iUP, iMAX) were increased at the three stimulus sites. Rheobase was increased (9.11 mA by TT, 9,77 mA by MM) and SDTC decreased (0,217 ms by TT, 0,224 by MM). The recovery cycle study showed a reduction of the absolute (MM) and relative refractory periods (TT and MM) without other striking abnormalities. Threshold electrotonus revealed a TEd(40-60) decrease and a current-threshold relationship steeper than in controls. Finally, median nerve ultrasound showed increased cross-sectional area at the wrist and elbow.
Conclusion:
This case of CMT1H could be considered a good model of hypertrophic demyelinating neuropathy, without secondary axonal loss. Most excitability

abnormalities are explained by an alteration of the passive properties of the membrane, with the exception of the decrease in SDTC which could be due to a lower density of persistent sodium channels, linked to demyelination.

Key words

nerve excitability measurements, CMT1H