ISOLATED DORSAL COLUMN AFFECTION IN LEBER´S HEREDITARY (OPTIC) NEUROPATHY 14484 (T-C), 4216 (T-C), 4917 (A-G) (CASE REPORT)

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A 44 year old female presented with tingling feet and fingertips. After exercise she felt exhausted, muscle stretch reflexes were brisk, 3 Hz repetitive and posttetanic stimulation was normal, VEP as visual acuity was normal. Tibial SEP N22 right 22,2 ms, left 22,6 ms, P40 right 51,3 ms, left 48,2 ms, where centrally prolonged. Right sural nerve ncv was 47 m/s. MRI of the head, cervical and thoracic spine was normal. MEP latencies to FDI and TA were normal, only the intensity to elicit MEP was higher than normal. Laboratory work up was unremarkable, including B12, copper, coeruloplasmin, dsDNS-ab, ANA, ANCA, long chain fatty acids, CSF, no ocb and no autochthonic immuno globulin synthesis. Only thyreoglobulin-ab were slightly increased 223 U/ml (115 U/ml), while TSH, fT3, T4, TPO were normal. She had been diagnosed with affected relatives (brother and cousin) to have a mutation at the locus 14484(T-C), 4216 (T-C), 4917(A-G) characteristic for hereditary Leber´s optic neuropathy, when she was 25y old. Her brother and her cousin had symptoms due to optic neuropathy, which she never had. The most probable cause of the patients dorsal column affection is hereditary Leber´s (optic) neuropathy without optic symptoms. When the mitochondrial dna locus 14484 is involved, affection of spinal cord columns in Leber´s optic neuropathy, had been reported, but there the optic nerve was affected too. The patient was tested during early adulthood for having this mutation, but developed atypical symptoms only later in her 40ies.