AN INTRIGUING CASE OF NEUROMUSCULAR DISEASE

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Introduction: FVL , masc, first consultation 04/10/2019, 45 yo. Symptoms: weakness in lower limbs for about 5 years, low back pain, intense fatigue when taking short walks. Physical exam: difficulty getting up when sitting, atrophy of thighs. FVC = 71%. **Exams results**: Muscle biopsy 12/2015 myopathic histological pattern with dystrophic features. . ENM 06/2016 myopathic pattern and signs of chronic denervation. Muscle biopsy 09/2019 histological pattern compatible with limb-girdle muscular dystrophy. PSG 10/2018: AHI = 19.4/h, minimum SpO2 = 90%, awakenings 24.4/h, presence of snoring, PLM = 4.2/h. Genetic Testing: Broad DNA Panel (Neuromuscular Diseases): Gene TRIM32 (Tripartite Motif Containing 32, OMIM* 602290, Position: chr9:116,699,528, Variation C > T, Consequence: p.Arg596Cys ENST00000450136), Copies: Homozygous (2 copies). **Diagnosis**: Girdle-type muscular dystrophy, autosomal recessive 8 (LGMDR8). **Discussion**: This variant is quite rare, being present in heterozygosis in 2 out of 125,000 individuals of the world's population and has never been previously described in the medical literature. It was submitted to the ClinVar variant repository and classified as of uncertain significance. Homozygous or compound heterozygous pathogenic variants in the TRIM32 gene are associated with autosomal recessive girdle-type DM 8 (LGMDR8, previously called LGMD2H), which has an age of onset in the first decade of life and a slow rate of progression. It leads to predominantly proximal muscle weakness and atrophy in the shoulder and pelvic girdles, myopathic gait, and elevation of creatine kinase. **Treatment:** L-Carnitine 1 gram 2 times a day, turmeric 500 mg 2 times a day, hydrotherapy 2 times a week. Currently, significant improvement of muscle pain and slow evolution of muscle atrophy

Key words: Girdle-type muscular dystrophy, TRIM32 gene, L-Carnitine treatment