

NAVIGATING THE COMPLEX LANDSCAPE OF MUSCULAR DYSTROPHY IN A PATIENT PRESENTED WITH PAIN

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Introduction

A 41-year-old man presented with several years of distressing pain, muscle fatigue, and twitching across his chest and proximal muscles. He was diagnosed with neuronal hyperexcitability, which further exacerbated his pain. Despite experiencing agonizing discomfort, his muscle strength appeared to be normal during the examination. However, the visible rippling in his proximal muscles added to his pain and concern. Extensive cardiac evaluation yielded no definitive findings.

Materials and Methods

As the case report is devoid of patient identifiable information, it is exempt from IRB review requirements as per Jefferson/Einstein policy.

Results

The results of the electromyography (EMG) were inconclusive, showing nonspecific myopathic features that added to the mystery of his pain. Meanwhile, his creatine kinase (CPK) levels were found to be elevated, reaching the 400's, which likely contributed to his ongoing pain and discomfort.

Interestingly, the journey extended to his family as well. His 9-year-old son had been diagnosed with muscular dystrophy, and two daughters also exhibited elevated CPK levels, all of which added to the family's concern.

Through genetic testing via Invitae muscular dystrophy testing, our patient received confirmation of a heterozygous C.80G>A(p.Arg27Gln) mutation in the Caveolin-3 gene. This missense mutation caused an amino acid alteration from Arg to Gln in the Caveolin 3 protein. The resulting decrease in Cav-3 protein expression in skeletal muscle tissue. This mutation followed an autosomal dominant inheritance pattern, potentially affecting future generations.

In light of these painful and challenging circumstances, it is essential to highlight the crucial role of pain management. While the design/methods of addressing this pain may not apply here, the results emphasized the broad spectrum of clinical presentations associated with mutations in the Caveolin-3 gene. These include limb girdle muscular dystrophy, rippling muscle disease, idiopathic persistent hyperCKemia, and distal myopathy. The complexity of these conditions magnifies the struggle of pain management and the necessity of timely diagnosis to alleviate the persistent pain and suffering endured by both patients and their families.

Discussion

In conclusion, muscular dystrophy, with its diverse manifestations, adds to the complexity of diagnosis and painful management. The awareness of clinical symptoms and a comprehensive family history become crucial tools for diagnosis, guiding healthcare professionals through the intricate landscape of painful phenotypic variations. The excruciating pain experienced by patients and their families underscores the importance of advancing research and treatments to address the various facets of pain associated with muscular dystrophy, including the profound impact on their quality of life.

References

1-A Role for Caveolin-3 in the Pathogenesis of Muscular Dystrophies Bhola Shankar Pradhan, Tomasz J. Prószyński Int. J. Mol. Sci. 2020, 21(22), 8736; <https://doi.org/10.3390/ijms21228736>