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Hereditary Inclusion Body Myopathy (HIBM) as a Rare Clinical Entity: A Case Report

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Introduction: Hereditary inclusion body myopathy (HIBM) is a rare slowly progressive muscle disease that frequently develops between age 20 and 40 years with bilateral foot drop induced by anterior tibialis weakness. Involvement of lower-extremity muscle continues from the anterior to the posterior compartment of the lower leg, followed by hamstrings, then hip girdle muscles, with correlative sparing of the quadriceps.

Methods: Description of the case report with hereditary inclusion body myopathy form in North Macedonia.

Results: The authors report female patient that is genetically homozygous of the pathogenic modification in the GNE (Glucosamine (UDP-N-Acetyl)-2-Epimerase/N-Acetylmannosamine Kinase) gene, which means that this condition is inherited from the both parents. Three years ago, the patient noticed instability in walking and peroneal type of walking, accompanied by weakness of the calf muscles. Furthermore, it is also important the fact that the patient's sister also was clinically diagnosed with this type of distal myopathy 16 years ago. In addition, the evolution of the disease in her is with progressive character matching with the weakness of the lower extremities in the earlier stage, together with the weakness of the upper extremities at a later stage.

Conclusion: A plenty of new facts remains to be learned in the future studies about the other and less common forms of HIBMs beyond the definition of the corresponding causative gene defect.