Muscle magnetic resonance imaging and histopathology in ACTA1-related congenital nemaline myopathy

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Introduction: Muscle biopsy is usually diagnostic in nemaline myopathy (NM), but some patients may show nonspecific findings, leading to pitfalls in diagnosis. Muscle MRI is a helpful complementary tool. Methods: We assessed the clinical, histopathological, MRI, and molecular findings in a 19-year-old patient with NM in whom 2 muscle biopsies with ultrastructural examination showed no nemaline bodies. We analyzed the degree and pattern of muscle MRI involvement of the entire body, including the tongue and pectoral muscles. Results: Muscle MRI abnormalities in sartorius, adductor magnus, and anterior compartment muscles of the leg suggested NM. A previously unreported fatty infiltration of the tongue was found. A third biopsy after the muscle MRI showed scant nemaline bodies. A novel heterozygous de novo ACTA1 c.611C>T/p.Thr204Ile mutation was detected. Conclusions: We highlight the contribution of muscle imaging in addressing the genetic diagnosis of ACTA1