17. A novel myopathy, two very different phenotypes: the French Canadian Experience.

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BACKGROUND: According to the modified Bohan and Peter classification, autoimmune myopathies with a phenotype of pure polymyositis (pPM), are defined by the absence of both overlap features and a dermatomyositis rash. However, many mimickers of autoimmune myopathies will have a pPM phenotype, and the diagnosis of a treatable myopathy is a challenge.

OBJECTIVE: Our objective is to report the first two French Canadian cases of a recently-described novel myopathy.

METHODOLOGY: We reviewed the clinical and pathological characteristics of two patients with an undiagnosed, and apparently unrelated, myopathy; the sequence of events that helped secure the diagnosis was analyzed.

RESULTS: A 44 year-old female presented with an unexplained liver problem. Over 11 years, three liver biopsies and three muscle biopsies failed to secure a diagnosis. The "pPM three steps to diagnosis" approach was used. A 21 year-old male presented with an acute myopathy. The same "pPM three steps to diagnosis approach" promptly confirmed the diagnosis.

CONCLUSION: Two French Canadian patients with a novel myopathy were diagnosed. The "pPM three steps to diagnosis" approach was instrumental to diagnose this entity.