Atypical clinical pictures and paucisymptomatic subjects: pitfalls in diagnosing inflammatory myopathies

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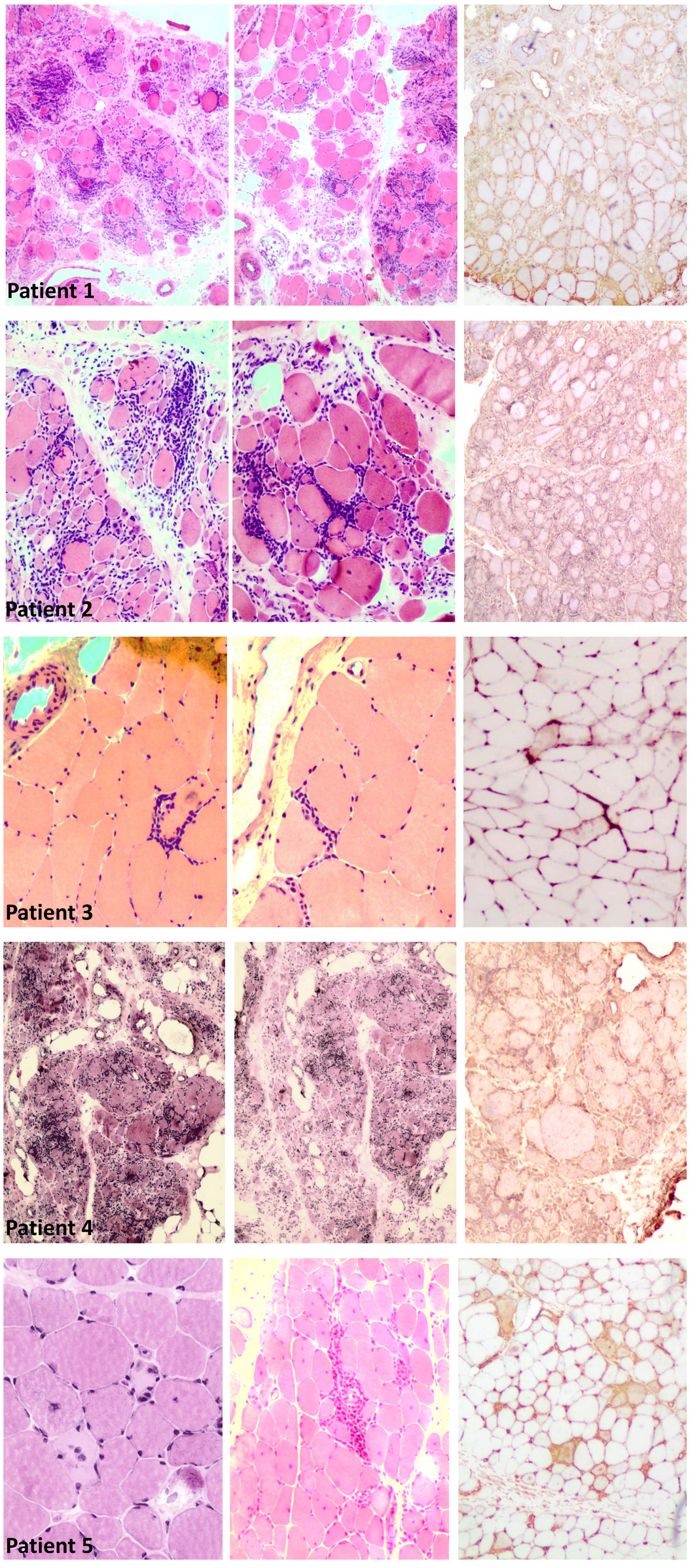
OBJECTIVES: To describe a small cohort of patients with history and clinical picture not typical for an idiopathic inflammatory myopathy (IIM) but having muscle biopsy features that meet the criteria for pathological diagnosis.

PATIENTS AND METHODS: Diagnostic muscle biopsy was performed in five patients. The first patient was a 67-year-old male complaining of exercise intolerance and proximal lower limb weakness since about ten years with no clinical progression. The second patient was a 73-year-old female who complained of "food bolus block" in the pharynx/esophagus since 7 years. A 66-year-old male patient complained of dysphagia, hypophonia and exercise-related dyspnea since two years without developing any significant limb and face muscle weakness. The fourth patient was a 65-year-old male who noted a progressive focal atrophy of the right thigh in the past five years; in the last year, proximal lower limb weakness and difficulty in rising from a squatting position and climbing stairs appeared. The fifth patient was a 72-year-old female which was submitted to muscle biopsy for the occasional evidence of increased levels of CK (2000 U/L). She did not complain of neurological symptoms. A personal and family history of neuromuscular disease, endocrinopathy, rheumatological diseases or exposure to myotoxic drugs or toxins was absent.



RESULTS: Muscle biopsy showed in all the patients pathological signs supporting diagnosis of inflammatory myopathies i.e. mononuclear cell infiltrates, muscle fiber necrosis, inflammatory cells surrounding, invading and destroying healthy muscle fibers and MHC class I antigen expression on fiber membranes.

CONCLUSIONS: Accepted criteria for diagnosing IIM include subacute and insidious development (usually over a period of 3-6 months) of limb symmetrical muscle weakness, most prominent in proximal muscles. A non-typical clinical picture may delay the diagnosis and the onset of proper therapy. Muscle tissue damage can be very severe despite the mildness of clinical picture and the diagnostic delay may affect the therapeutic response.

Our study warns that atypical clinical pictures can be easily found in patients with IIM and muscle biopsy can be critical for obtaining diagnosis and ensuring the proper treatment.