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A rare disease presenting with severe hyper CKemia: the antisynthetase syndrome

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Background: Different pathological conditions may present with elevated serum creatine kinase (CK), a condition known as hyperCKemia. Common causes include myocardial infarction, neuromuscular disease, cholesterol-lowering medication, physical exercise.

Clinical case description: A 59-year-old woman attended the emergency department with sudden dyspnea and four weeks-lasting proximal muscle weakness. CK levels were very high (>18.000 U/L). She reported no other symptoms or significant past history. We then saw her at our inpatient Internal Medicine clinic and found that she also had a history of symmetrical hand arthritis, acrocyanosis with hyperkeratotic lesions at index fingers and feet. Routine investigations showed increased CK (15770 U/L), LDH (1897 U/L), GOT (1012 U/L), GPT (622 U/L), CRP (12.1

mg/L) levels. Tests for antinuclear (ANA) and anti-Jo-1 antibodies were found positive. Electromyography and magnetic resonance imaging confirmed muscle disease. Chest high resolution CT scan showed interstitial lung disease (ILD). The patient was diagnosed with antisynthetase syndrome and started on high dose steroid and mycophenolate mofetil. Given the mild improvement, two weeks later treatment with high dose i.v. immunoglobulins was added.

Conclusions: Antisynthetase syndrome is an autoimmune disease characterized by ILD, myositis, arthritis, and specific antibodies, most commonly anti-Jo-1. Here we underline the relevance of suspecting idiopathic inflammatory myopathy, including antisynthetase syndrome, in individuals presenting with unexplained raised CK levels.