A Mystery Case of Incidental Transaminitis in the Setting of Rheumatologic Disease

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Introduction: Transaminitis, characterized by elevated liver transaminases, can stem from numerous underlying causes, posing challenges in diagnosis and management. In this case, we describe a patient case with unclear etiology amidst multiple chronic conditions and nonspecific symptoms.

Case Presentation: A 66-year-old Caucasian female presented to the ED after a fall, with symptoms of increased somnolence, forgetfulness, and weakness over months. Her medical history includes CHF, COPD, GERD, pulmonary hypertension, and uveitis. Initial lab results showed elevated AST (1994 U/L), ALT (780 U/L), and ALP (195 U/L). Imaging was negative. Vital signs included elevated BP and low oxygen saturation. She reported mild chest pain and dyspnea due to CHF exacerbation, and later developed abdominal pain. Her history included intermittent elevated transaminases, swollen fingers, peripheral numbness, arthralgias, and dysphagia, with a past positive anti-RNP titer. Differential diagnoses included drug-induced toxicity, autoimmune hepatitis, infectious hepatitis, and ischemic shock liver. Testing revealed positive anti-RNP, ANA, p-ANCA antibodies, and low C4. She was treated for CHF with steroids, leading to stabilization and down-trending LFTs. Suspected of having MCTD, she was referred to outpatient rheumatology.

Conclusion: Mixed Connective Tissue Disease (MCTD) is a rare condition characterized by a mix of symptoms from systemic sclerosis, systemic lupus erythematosus, and polymyositis, with high-titer anti-U1-RNP antibodies. Diagnosis is difficult due to nonspecific symptoms and varying criteria. A 2020 study found the Kasukawa criteria most sensitive, requiring one common symptom (e.g., Raynaud's phenomenon), positive anti-RNP antibodies, and signs of at least two mixed syndromes. Although autoimmune hepatitis is rare in MCTD, it should be considered in patients with unexplained liver issues.

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This case highlights the importance of considering autoimmune causes like MCTD in complex transaminitis cases, highlighting the need for a thorough patient history and differential diagnosis.

Keywords: Transaminitis, Mixed Connective Tissue Disease, MCTD, Autoimmune Hepatitis, Chronic Condition, Elevated Liver Transaminases, Anti-RNP Antibodies

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