中文題目：MDA5 抗體陽性皮肌炎併發快速進展間質性肺病之病例報告

英文題目：Fatal Rapidly Progressive Interstitial Lung Disease in Anti-MDA5 positive Dermatomyositis: A Case Report

作　　者：李宜霖[1], 杜漢祥[2*], 古關光浩[1], 林永章[1], 廖家德[1], 許向平[3]

服務單位：[1]奇美醫院內科部, [2]奇美醫院加護醫學部, [3]奇美醫院外科部

Introduction: Anti-MDA5 positive dermatomyositis has a high risk of developing rapidly progressive interstitial lung disease (RPILD), which causes poor prognosis and a high mortality rate. Early diagnosis and treatment are crucial. Here, we report a patient with fatal RPILD refractory to medical therapy.

Case presentation: A 32-year-old woman with newly diagnosed diabetes mellitus presented with two weeks of intermittent fever, skin rashes and dyspnea. Typical findings of V sign, Heliotrope sign, Gottron papules and periungual erythema confirmed the diagnosis of dermatomyositis but without evidence of myositis. Respiratory failure developed soon, and exams yielded pulmonary aspergillosis and Legionellosis. Intermittent fever was noticed despite antibiotics and steroid therapy, and deteriorating hypoxemia happened in vain of mechanical ventilation support. Chest CT revealed subpleural atelectasis of lower lungs with traction bronchiectasis, and bilateral hilar ground glass opacity. Autoimmune profiles showed negative ANA, negative anti-Ro & anti-La, but weak positive anti-MDA5 antibody. A diagnosis of clinically amyopathic dermatomyositis (CADM) with anti-MDA5 antibody-associated RPILD was impressed. Immunosuppressant combination therapy was prescribed, in addition to methylprednisolone pulse therapy, rituximab, and plasma exchange. Although mild improvement in oxygenation was noticed initially, her clinical condition kept deteriorating despite those intensive treatments. The patient died from progressive hypoxemia and hypercapnia under VV-ECMO support.

Discussion: The diagnosis of connective tissue disease-associated interstitial lung disease mainly depends on clinical suspicion, imaging study, immunological profile, and pathology. Non-specific interstitial pneumonia, which mainly presented with basal predominant reticular abnormalities in the lower lung zone with traction bronchiectasis and ground-glass opacity, is commonly seen in imaging studies. In our case, the patient presented initially with signs and symptoms of pneumonia, which might disguise the underlying RPILD and make the diagnosis more complicated. It is essential to remind the atypical presentation of CADM compared to dermatomyositis, including the lack of myositis and a negative ANA titer. RPILD is highly associated with anti-MDA5 antibody in such cases, and thus, early intensive combined immunosuppressive therapy is crucial as the treatment. According to the literature, combining medications with glucocorticoids, calcineurin antagonists, and cyclophosphamide are the favourable first-line treatment. Rituximab, plasma exchange, and IL-6 receptor antagonists are commonly used as an add-on or salvage therapy for those refractory cases. Even though, the overall mortality of the case exceeds 50% in the review.

Conclusion: Dermatomyositis patients with clinical and imaging findings of interstitial pneumonitis should prompt the diagnosis of dermato-pulmonary syndrome. Anti-MDA5 antibody should be checked, although the exam is not available widely. Due to the high risk of developing RPILD, intensive combined immunosuppressants given early in the disease course might improve overall morbidity and mortality.