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NSIP secondary to Anti-synthetase syndrome

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Dear Editor,

Shortness of breath is one of many common presenting complaints to the respiratory clinic, this single symptom could be caused by a large range of medical conditions. We report the case of a 54 year old lady that was referred to our clinic with worsening shortness of breath on exertion and a dry cough. HRCT thorax was performed and demonstrated sub-pleural based bi-basal fibrosis with traction bronchiectasis. ANA, ENA were positive and extended myositis panel revealed positive autoantibodies; Mi2 beta , PL-12 and Ro52. A diagnosis of Anti-synthetase syndrome was reached and she was commenced on mycophenolate and prednisolone. Symptoms were much improved with this therapy. This case highlights the importance of a detailed clinical history and subsequent relevant investigations to ensure the early correct diagnosis and treatment of rare medical conditions.

Confirming a diagnosis of a rare condition can be quite challenging, we are always taught 'common is common', and this is generally true. A rare disease is defined as a condition which affects less than 1 in 2,000 people. Current estimations have revealed over 7,000 rare diseases, and this number is only increasing with the advances in medical research.[1] Going back to the basics of our training, history and clinical examination, these are the fundamental skills needed to identify the rarities in a vastly common medical world.

A 54 year old lady MD was referred to our respiratory clinic from her GP with worsening SOB, the GP had been treating her as asthma. On further questioning she revealed her exercise tolerance had significantly reduced over a period of 2 months, currently can only walk 50 yards before needing to stop due to dyspnoea, previously able to walk approximately 5kms 3 times a week with no issue. Physical examination in clinic was largely unremarkable. MD was trialled on therapy with Seretide by the GP, which did not result in any improvement. The GP did arrange a CT Thorax which showed sub pleural reticulation with fibrotic changes associated with traction bronchiectasis. Following our initial detailed clinical history, physical examination and imaging results, we further investigated with blood tests and PFTS. Initial PFTs showed a mild to moderate restriction, subsequent PFTs showed a severely decreased DLCO of 31%. ILD bloods were sent which revealed positive autoantibodies; Mi2 beta [2], PL-12[3] and Ro52[4], the rest of the blood screen was negative. The above autoantibodies in conjunction with the clinical findings and CT results was suggestive of a diagnosis of anti-

synthetase syndrome. We involved our Rheumatology colleagues who commenced MD on Mycophenolate and Prednisolone, her dyspnoea has much improved.

Anti-synthetase syndrome is a rare and complex auto-immune disease, which is a subset of the idiopathic inflammatory myopathies[5], with an incidence of 0.56 per 100,000 and prevalence of 9 per 100,000.[6]

There are two main diagnostic criteria used for anti-synthetase syndrome; Connors et al proposed that for a diagnosis the patient required the presence of an anti- aminoacyl tRNA synthetase antibody plus at least one of the following clinical features; Raynaud's, arthritis, ILD, Fever and Mechanic's hands, Solomon *et al* proposed alternative, stricter criteria, requiring two major or one major and two minor criteria, in addition to the presence of an aminoacyl-tRNA synthetase autoantibody.[7] The most common being anti- Jo-1, and one of the less commonly seen anti-synthetase antibody is the ant-PL-12, as seen in our case.[7]

Corticosteroids are considered the first line therapy, however when used as monotherapy there is frequent relapse of the lung disease when attempting to taper the steroids.[7] Therefore, as in our case, a steroid-sparing agent (usually mycophenolate or azathioprine) can be used along with the steroid and has shown a better survival and fewer relapses in comparison to steroid monotherapy.[8]

This case highlights the importance of a detailed clinical history and physical examination, followed by relevant investigations to ensure the early correct diagnosis and subsequent treatment of rare medical conditions.

Declarations of Conflicts of Interest:
None declared.

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