Case Report

An unusual presentation of Mixed Connective Tissue Disease: a case of chronic pulmonary thromboembolism and Nephrotic range proteinuria

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Background

Mixed connective tissue disease (MCTD) is a rare autoimmune rheumatological disease which mostly affected in young females. It has a combination of symptoms and signs of Systemic Lupus Erythematosus, Systemic sclerosis, Rheumatoid arthritis and myositis along with high titer of U1RNP. Chronic pulmonary thromboembolism is a rare manifestation in MCTD.

Summary

A 31 year old Sri Lankan female presented to us at the age of 24 year with rheumatological manifestations. She initially presented with bilateral symmetrical polyarthritis and finger Raynaud`s. She was managed as seropositive rheumatoid arthritis. Following years she gradually developed dyspnoea, investigations were confirmed pulmonary hypertension later Nonspecific interstitial pattern of ILD. Her CTPA was negative but ventilation perfusion scan became positive for ventilation-perfusion mismatch and confirmed chronic pulmonary thromboembolism. Her thrombophilia screening was negative. Over the years she developed features of MCTD.

At the age of 31 year old, she developed severe Raynaud`s leading to necrotic right big toe and nephrotic range proteinuria. It is again a rare manifestation in MCTD. ESR -100mm/hr, UACR-3.5, SCL70 negative,anti-Centromere antibody negative , DsDNA-negative,anti-U1RNP antibody > 10000U/ml .

She was initially on Methotrexate with Nifedipine, later it was changed to Azathioprine considering ILD. Furthermore at the final state she was given intravenous steroid pulses followed by MMF. Bosentan also added due to severe Raynaud`s. She gained a remarkable response in all the aspects including recovery of proteinuria.

Conclusion

This case highlights a rare presentation of MCTD with chronic pulmonary thromboembolism and nephrotic range proteinuria. Pulmonary hypertension is an early manifestation and Raynaud`s is a well-known feature in the early disease course. Though absence of severe renal or central nervous system involvement is a hall mark of MCTD, severe proteinuria has been reported mainly due to membranous nephropathy.

Keywords

MCTD, Nephrotic range proteinuria, chronic pulmonary thromboembolism, Bosentan