MIXED CONNECTIVE TISSUE DISEASE COMPLETES 50 YEARS – ANALYSIS OF A SERIES OF 161 PATIENTS

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BACKGROUND

Since its initial description in 1972, mixed connective tissue disease (MCTD) has been the subject of numerous controversies. This study analyzes a series of patients with MCTD followed at a single tertiary referral service.

METHODS

This retrospective study (2019–2021) analyzed patients with a clinical diagnosis of MCTD who presented positive anti-RNP, initially by hemagglutination (> 1:1,000) and later by ELISA, in the absence of other specific autoantibodies. The four proposed sets of classification criteria (Sharp, Kahn, Kasukawa and Alarcon–Segovia) were applied. Demographic, clinical and laboratory criteria were analyzed, as well as the main disease categories: systemic sclerosis (SSc), systemic lupus erythematosus (SLE) and inflammatory myopathy.

RESULTS

Among the 161 patients included, most were female (93.78%) and white (61.5%). The mean age at diagnosis was 39 years, with a mean follow-up of 12 years. One hundred forty-one (87.6%) patients met at least 1 set of criteria, with 12 (7.5%) meeting 1 set, 33 (20.5%) 2 sets, 12 (7.5%) 3 sets and 84 (52.2%) all 4 sets. The other 20 patients had symptoms of at least 2 disease categories, but did not meet any classification criteria for MCTD. All patients had Raynaud's phenomenon. One hundred seventeen (72.7%) patients were considered to have an SSc pole, with the most frequent clinical manifestations being esophageal dysmotility (85.1%), puffy fingers (55.3%), sclerodactyly (27.3%), nail pitting scars (25.5%) and telangiectasias (19.3%). Interstitial lung disease was present in 48.4% of the patients, while pulmonary hypertension was observed in 16.1%. The SD pattern on nailfold capillaroscopy (NFC) was present in 59% of patients, with only 13% with normal NFC. A myopathy pole was observed in 98 (60.8%) patients, who complained of muscle weakness and had increased creatine phosphokinase levels. One hundred thirty-nine (86.3%) patients had SLE pole, with the main findings being: leukopenia (75.2%), facial erythema with photosensitivity (23.6%), thrombocytopenia (19.3%) and serositis (15.5%). Arthritis or inflammatory arthralgia were reported by 84.5% of patients. Treatment included glucocorticoids (90.1%, with a dose higher than 15 mg in 65.2%), azathioprine (72.1%), methotrexate (63.9%), cyclophosphamide (39.1%) and leflunomide (26.7%). To control Raynaud's phenomenon and digital ulcers, 88.2% received calcium channel blockers and 11.2% received sildenafil. For esophageal symptoms, 96.3% received proton pump inhibitors and 75.8% received prokinetics.

CONCLUSION

Despite the controversies, the careful application of the MCTD classification criteria, in the presence of anti-RNP and in the absence of other specific autoantibodies, allows the diagnosis and consequent treatment of a considerable number of patients.

KEYWORDS

Mixed connective tissue disease, Anti-RNP, Raynaud's phenomenon, Systemic sclerosis, Systemic lupus erythematosus.