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CASE REPORT Open Access

Mixed connective tissue disease: presenting as trigeminal neuralgia

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Abstract

Connective tissue diseases (autoimmune rheumatic diseases) are a group of disorders of unknown etiology. The term mixed connective tissue (MCTD) is used when two or more autoimmune diseases occur in the same individual simultaneously, for example, systemic lupus erythematosus, scleroderma, polymyositis, dermatomyositis in various combinations. Here, we report a case of MCTD presenting as trigeminal neuralgia.

Introduction

Mixed connective tissue disease is a rare autoimmune disorder that has 3 other connective tissue diseases (systemic lupus erythematosus, scleroderma, and polymyositis) along with the presence of a distinctive antibody U1-ribonucleoprotein (RNP) [1, 2].

We report a case of MCTD presenting as trigeminal neuralgia.

Case

Thirty-eight-year-old female with no known comorbidities presented pain over the right side of the face and scalp for 4 months, severe paroxysmal pain lasting for approximately less than 2 min, and aggravated with chewing. Later on, she developed tightness of the skin of the lower limbs progressing to the upper limbs and face associated with hardening of the skin. For the past 2 months, she had difficulty getting up from a squatting position.

On examination

Fixed mouth opening (Fig. 1), hard thick non-pinchable skin, hypopigmented patches with perifollicular pigmentation over her neck shoulders and chest, and sclerodactyly were present. The nervous system exam showed reduced sensory perception in the area supplied by V_2 division of the trigeminal nerve and minimal proximal muscle weakness with power of grade 4. Respiratory system bilateral fine crepitation's. Other systems are within normal limits.

Investigations

HB 11.6g/dl, total count 9170 cells/cumm, platelet 5.52 lakhs/cumm ESR 46

Total biliribuin 0.6, AST-188, ALT 135, ALP-7

Na+ 133 meq/L, K+ 4.1 meq/L, RA factor 8.6

Total CPK 4106 U/L, ANA speckled pattern, ANA titer 1:1000, ANA profile U1rnp, strongly positive

Chest X-ray: right descending pulmonary artery prominent (Fig. 2), HRCT suggestive of interstitial lung disease. Echocardiography showed no RWMA, good LV function, and mild pulmonary artery hypertension.

Management

Started on T.PREDNISOLONE 30 mg OD

T. carbamazepine 200 mg OD for trigeminal neuralgia

Improved symptomatically after the 1 week of admission and advised regular follow-up and started tapering steroids.

Discussion

MCTD is a rare autoimmune disease, in which one disease overlaps with the other. U1rnp antibody is specific to MCTD [3]. MCTD can affect people of all ages, and it is most commonly seen in women under the age of 30 [4].

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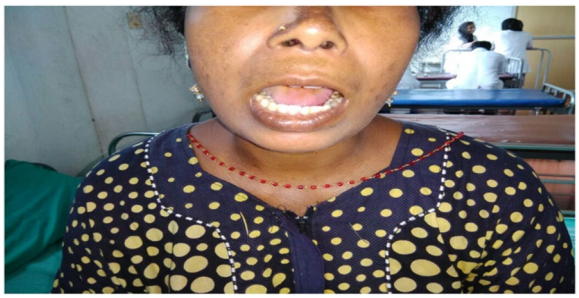

Fig. 1 Fixed mouth opening

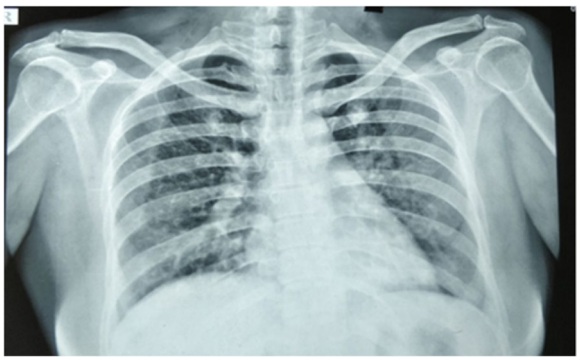

Fig. 2 Chest X-ray: right descending pulmonary artery prominent

The exact underlying cause of mixed connective tissue disease (MCTD) and the incidence is not known. Signs and symptoms may include arthritis, Raynaud's phenomenon, myocarditis, interstitial lung disease, skin abnormalities, muscle weakness, and esophageal dysmotility [5]. MCTD is often suspected after a physical examination reveals signs and symptoms associated with the condition. The diagnosis is supported by U1 RNP positivity [6].

Our case's initial symptoms were pain over the right side of the face and scalp which was worsening in cold air, chewing suggestive of trigeminal neuralgia, and later on developed tightness of the skin and non-pinchable skin on the bilateral upper limbs and sclerodactyly. She developed difficulty in getting up from a squatting position with high CPK levels indicative of polymyositis. The lack of CNS involvement is characteristic of MCTD, but mild involvement has been described in about 25% of patients [7]. Out of which, trigeminal neuralgia was the most common neurological manifestations [8]. Chest X-ray was suggestive of pulmonary arterial hypertension. Hence, we suspected a MCTD and did an ANA profile which came out to be strongly positive for U1RNP. Trigeminal neuralgia can be presenting manifestations of MCTD. So, a high index of suspicion is required to make the diagnosis.

Conclusion

We report a case of a rare presentation of rare disease (MCTD) for the awareness of physicians and help them to make an accurate diagnosis which is very much helpful for the patient because MCTD has better prognosis compared to individual diseases.

Authors' contributions
The author(s) read and approved the final manuscript.

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Declarations

Ethics approval and consent to participate
We certify that we have obtained all consent forms from the patient.

Competing interests
The authors declare that they have no competing interests.

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