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CASE REPORT: MIXED CONNECTIVE TISSUE DISEASE (MCTD)

Hirni J Patel — Pharm D Intern, Parul Institute of Pharmacy and Research, Parul University, Vadodara, Gujarat.

Kuldip Panchal — Pharm D Intern, Parul Institute of Pharmacy and Research, Parul University, Vadodara, Gujarat.

Dhrupesh N Ghatodiya — Pharm D Student, Parul Institute of Pharmacy and Research, Parul University, Vadodara, Gujarat.

S P Srinivas Nayak* — Assistant Professor, Department of Pharmacy Practice, Parul Institute of Pharmacy and Research, Parul University, Vadodara, Gujarat. *Corresponding Author

Mohit Buddhadev — Assistant Professor, Department of Pharmacy Practice, Parul Institute of Pharmacy and Research, Parul University, Vadodara, Gujarat.

G.S. Chakraborthy — Professor and Principal, Parul Institute of Pharmacy and Research, Parul University, Vadodara, Gujarat.

ABSTRACT

Background: The term mixed connective tissue disease (MCTD) was coined by Sharp and colleagues in 1972 proving to that mixed connective tissue disease a systemic autoimmune disease which involves antibodies like anti-sm, Ro-52 recombinant, nucleosome, anti-Jo-1, anti ds-DNA, etc.

Case presentation: A 28-year-old lady presented with complaints of multiple joint pain since 3 days along with weakness. She also complained about low grade fever associated with chills and rigor, vomiting, headache and body ache. There was involvement of morning stiffness in small joints of fingers and toes. Reports were done and she was eventually treated according to it.

Conclusion: The work has reported a rare case of an adult patient with MCTD characterized by a severe multiple joint pain along with severe weakness and positive AntiU1-RNP levels and was treated with corticosteroid (Prednisolone), Methotrexate and analagesic (Tramadol and Paracetamol).

INTRODUCTION:

The term mixed connective tissue disease (MCTD) was coined by Sharp and colleagues in the year 1972 to describe a class of patients who had overlapping clinical presenations of systemic lupus erythematosus (SLE), scleroderma, and polymyositis (PM) [1]. Mixed Connective Tissue Disease (MCTD) is characterized by the presence of auto antibodies and T cells reactive with U1 ribo-nucleoprotein (U1-RNP) polypeptides of the spliceosome complex including their associated uridine-rich (U) small nuclear RNAs immuninologically. Hence it is a systemic autoimmune disease [2, 3]. More studies have showed that anti-sm antibodies are restricted to patients with SLE. MCTD syndrome is most compatible with SLE modified with presence of RNP antibodies, since it's not accepted as different clinical entity [4, 5]. Arthralgias and arthritis is the commonest clinical presentation of mctd and our patient was treated for it with analgesics. As pain increased along with other symptoms of mcdt like polymysitis, joint pain [6]. Initial descriptions of this syndrome emphasized a good prognosis with an excellent response to corticosteroid therapy like oral Prednisolone [6, 7].

CASE REPORT:

A 28-year-old lady presented with complaints of multiple joint pain since 3 days along with weakness. She also complained about low grade fever associated with chills and rigor, vomiting, headache and body ache. There was involvement of morning stiffness in small joints of fingers and toes. She had been given steroids and analgesics for the treatment. On asking, these symptoms had been worsening over a period of time. There was no history of skin rashes, abdominal pain, and pedal oedema. The patient is a known case of diabetic and not on regular medications. There was no family history of diabetes mellitus, hypertension, etc. On examination, it was revealed that the patient had pallor. The fingers showed spindle shaped deformity but there was swelling. But there was presence of finger blackening in right hand, and presence of blackening in left lower limb. Her blood pressure was 100/90mmHg, pulse was 88/min. During systemic examination, power in muscles was 5/5 and reflexes were normal, all other systems were normal. A provisional diagnosis of mctd varied with rheumatoid arthritis, systemic lupus erthematosis was further investigated.

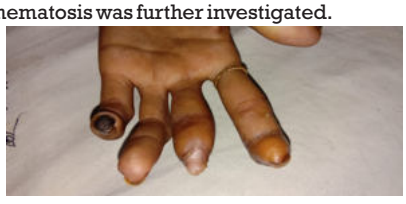

Figure.1. Sclerodactyly, Raynauld's amputation along with swelling of fingers

Lower limb angiography revealed moderate to significant stenosis involving right and left PTA and peroneal trunk with diffuse non calcified thromboembolism, mild stenosis involving left ATA with diffuse non calcified thromboem bolism.

Table 1:- Ana Profile (immune Dot-autoimmunity Screening) With Intensity Of ANA

TEST/	RESULTS	Intensity	Class
ANA IF	Coarse speckled, cytoplasm homogenous ++, positive	-	-
Positive control	Positive +++	75	+++
RNP/Sm	Positive +++	101	+++
Sm	Positive +	22	+
Ro-52	Positive +++	56	+++
Scl-70	Negative	1	0

Centromere B	Negative	0	0
Nucleosomes	Positive +++	64	+++
AMA-M2	Negative	1	0
Histones	Negative	1	0
Anti-Jo-1	Negative	2	0
Anti-Sm	Negative	1	0
Anti ds-DNA	Negative	0	0
anti-centromere	Negative	0	0
Rib- P Protein autoantibodies	Negative	1	0

Explanation

INTENSITY	CLASS	EXPLANATION
0-5	0	Negative
6-10	(+)	Borderline
11-25	+	Positive
26-50	++	Strong positive
51-256	+++	Strong positive

Table 1.2:- Ana Report Interpretation

AUTO ANTIBODY	COMMENTS
RNP/Sm	• Mixed Connective Tissue Disease (MCTD) 95-100% • Disseminated Lupus Erythematous 30-40%
Sm	• Disseminated Lupus Erythematous 30-40%
Ro-52	• Scleroderma • Rheumatic disease • Myositis
Scl-70	• Progressive Systemic Sclerosis 25-75%
Jo-1	• Polymyositis 25-35%
Ds-DNA	• Systemic Lupus Erythematous 40-90%
Nucleosomes	• Systemic Lupus Erythematous
Histones	• Disseminated Lupus Erythematous 30-70%
Rib- P Protein	• Systemic Lupus Erythematous • Sharp Syndrome
AMA-M2	• Primary Biliary Cirrhosis • Chronic Liver Disease 30% • Progressive Systemic Sclerosis 7-25%

Other tests like hepatitis, HIV, Dengue, Chinkunguya all were negative. Therefore, a final diagnosis of Mixed Connective Tissue Disease was made.

Table 1.3:-Treatment plan

Sr. No	DRUG	DOSE	FREQ
1	INJ PARACETAMOL	1A (500mg)	SOS
2	INJ MULTIVITAMIN WITH VITAMIN B12	1A	1-0-1
3	T. TRAMADOL	50mg	1-0-1
4	T. METHYLPREDNISOLONE	5mg	1-0-1
5	T. METHOTREXATE 7.5MG OD	7.5mg	1-0-0
6	INJ CEFTRIAXONE	1gm IN 100ML NS	1-0-1
7	INJ PANTOP	1A (40mg)	1-0-1
8	INJ EMSET	1A (4mg)	1-1-1
9	TAB FOLIC ACID	5mg	1-0-0
10	TAB NIFEDIPINE	20mg	1-0-0
11	TAB ASPIRIN	75mg	0-1-0

There was plan for auto amputation when parents come for follow up after 1.5 months.

DISCUSSION

In the present study, we have described a rare case of female patient with MCTD who presented initially with severe multiple joint pain and weakness. Around 60% of patients have inflammatory myositis present with subclinical myopathy as their complaints. However, one can achieve quick clinical and laboratory response with low dose corticosteroids [8, 9]. Vasculitic neuropathy can be associated as distal symmetric neuropathy as they come together. Compressional neuropathies like carpal tunnel syndrome can be seen and hence neuropathy often causes difficulty in the diagnosis of mixed connective tissue disorder. Our patient presented with severe Raynaud's amputation and gangrene [10]. SLE is also associated with the 'markers', anti-double-stranded (ds) DNA or anti-Sm. However, antibody types do not always respect tradition: many coincide with parts of the spectrum in which diseases overlap with each other. The best known example is the antibody that reacts with U1 ribo-nucleoprotein (RNP), which is found in patients with overlapping features of SLE, systemic sclerosis and poly-myositis which has been termed mixed connective tissue disease MCTD. MCTD could be distinguished from SLE on the basis that SLE sera reacted with the RNase-resistant component [4]. Our patient had fulfil the criteria for the MCTD diagnosis. Thus, we concluded that it was confirmed case of MCTD. MCTD has no specific treatment due invlovment of multi organs but studies have proved that use of corticosteroids are very useful and other treatment could be done according to the symptoms which patient presents with like Nifedipine, Analgesics, Tumor necrosis factor blockers like Adalimumab, Infliximab, Anti-diabetic drugs.

CONCLUSION

This work has reported a rare case of an adult patient with MCTD characterized by a severe multiple joint pain along with severe weakness and positive AntiU1-RNP levels at presentation that was treated after the administration of corticosteroids (Prednisolone), and Methotrexate and then maintaining at low dose of corticosteroids along with analgesics like Tramadol plus Acetaminophen Combination drug.

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