Original Research Paper

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Rheumatology

A RARE CASE OF MIXED CONNECTIVE TISSUE DISORDER IN A MALE PATIENT

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ABSTRACT **INTRODUCTION :** Mixed connective tissue disorder{MCTD}, also known as Sharp's syndrome is a rare disease with female : male preponderance 16:1. it is rarely reported in india. MCTD is a disease with overlapping features of many connective disorders with anti U1 RNP positivity.

We present a male patient with fever,skin rash,painful joint swellings as initial presentation that led to diagnosis of MCTD.

CASE REPORT : A 40 year old male working as farmer presented with initial complaints of fever,joint sweling and pains sincce 15 days.physical examination revealed diffuse erythematous rash over the body,malar rash over the face,swollen fingers with severe small joint tenderness.after excluding all cause of fever with rash, laboratory workup showed MCTD.

Many criteria have been described to classify MTCD.our patient satisfied kasukawa diagnostic criteria.

KEYWORDS : MCTD ; mixed connective tissue disorder,anti U1 RNP antibody.

INTRODUCTION

Mixed connective tissue disease [MCTD] is an autoimmune disease first described in 1972 by sharp et al as a disease syndrome with overlapping features of systemic sclerosis,systemic lupus erythematosus [SLE] and polymyositis. Therefore it is referred to as an overlap disease.

The initial presentation of patients usually comprises non specific signs such as swollen hands,arthralgias,muscle weakness,Raynaud's phenomenon,shortness of breath on activity,fatigue.

Over a time the symptoms are dominated by smptoms of either of one of three illness along with high titres of anti U1 RNP antibody.

The etiology of the mixed connective tissue disorder is unknown but being autoimmune disease can run in families and known to effect women more than men.It is more common in females with a sex ratio of 16:1.No casual association and the varied presentation makes the diagnosis of this rare condition difficult.

We encountered one case presenting with fever ,generalised rash,swollen digits as initial presentation leaading to diagnosis of Mixed connective tissue disorder.

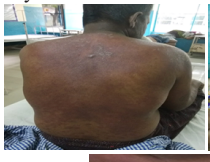
CASE REPORT

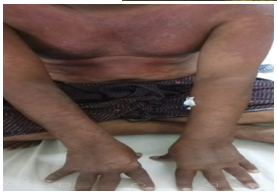
A 40 year old male patient presented with complaints of fever with generalised rash all over the body,severe myalgias since 15 days.he had a past history of small joint pains since 6 months.he also had history of recurrent oral ulcers and rash over the face.

Family history is nil significant.

On examination patient is found to have diffuse erythematous rash over the body,malar rash over face and swollen digits with tenderness,no oral ulcer at the time of examination

Erythematous rash





Swollen hands

On investigating the patient,thrombocytopenia with platelet count of 60,000,leukopenia was seen, liver and renal function tests are within normal limits malaria kit for antigen detection,dengue serology,leptospira antibodies,weilfelix test were negative.

Slit skin examinatin for lepra bacilli was also negative.

Complete urine examination,chest X- ray,2 dimensional echocardiogram – was normal.

ANA with immunoflourscence was strongly positive,ANA profile was positive for anti U1 RNP,anti histone antibodies,anti Ro antibodies were also positive.

Serum Creatine phosphokinase levels are raised – 250IU.

Based on above findings diagnosis of Mixed connective tissue disorder was made based on kasukawa criteria.

Kasukawa's Diagnostic Criteria

Must have One finding in Two of these Categories

Raynaud's or Swollen Fingers / Hands	
Anti-RNP antibodies	
SLE Signs or Symptoms	
	Polyarthritis
	Facial Rash
	Serositis
	Lymphadenopathy
	Leukopenia
	Thrombocytopenia
Scleroderma Signs or Symptoms	
	Sclerodactyly
	Pulmonary Fibrosis
	Vital Capacity <80%
	CO Diffusion <70%
	Decreased Esophageal Motility
Dermatomyositis Signs or Symptoms	
	Muscle Weakness
	Increased CK
	EMG Abnormalities

Patient was treated with oral steroids prednisole 60mg initailly and dose was tapered, after normal fundus examination hydroxychloroquine 200mg PO per day was started.

Patient symptoms improved with the above treatment.

DISCUSSION :

MCTD is a rare disease with unknown etiolgy,few cases have been reported after occupation of vinyl chloride,even after breast augmentation surgeries.there are familial cases with increased instance of HLADR4 compared with controls.

MCTD should be kept as differential diagnosis when overlapping signs of autoimmune diseases are present such as malar rash,arthralgia,myalgia,generalised rash,fatigue and generalised malaise.

Many criteria have been described to classify MCTD. Alarcon-segovia's criteria,kasukawa diagnostic criteria for MCTD.

Our patient satisfied kasukawa criteria with common symptoms of swollen hands, presence of anti U1 RNP,mixed findings of thrombocytopenia,leucopenia,elevated CPK values.

The therapy for MCTD would combine a cocktail of drugs to supress inflammation including NSAIDS like naproxen,cox-2 inhibitors like celecoxib,steroids such as prednisone, antimalarials like hydroxychloroquine,immunosupressants like azathioprine.Raynauds phenomenon can be treated with nifedipine,nitroglycerine.TNF blockers are etanercept and TNF antibodies infliximab,adalimumab in inflammatory arthritis.

The use of therapeutic antibodies like rituximab,a CD 20 receptor blocker in MCTD have shown benefit in cases of refractory polymyositis.

CONCLUSION :

Mixed connective tissue disorder is less common in males than females with increased risk of complications and mortality when compared with female counterparts.