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A RARE CASE REPORT OF MULTIPLE CONNECTIVE TISSUE DISEASE WITH SEVERE PULMONARY HYPERTENSION

General Medicine

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ABSTRACT

A 38 year old female presented with complaints of tightness, dryness, hyperpigmentation of skin , tightness of mouth, tingling numbness and pain in fingers while handling cold water since 6 months. Exertional breathelessness since 1 month .She had history of swelling of hands six months back. Finger examination showed sclerodactyly and ulceration over skin on interphalangeal joints of both hands without any deformity .Since clinical findings were suggestive of Raynaud's phenomenon, sclerodactyly, swelling of hands , scleroderma, she was further investigated for presence of connective tissue disease . On immunological investigations , ANA profile was positive with presence of strong positive U1RNP antibodies (titre>1:1600). Chest x ray and 2d echocardiography revealed presence of Pulmonary Hypertension. She was diagnosed with Multiple Connective Tissue Disease (MCTD) with severe pulmonary hypertension based on clinical , radiological and laboratory findings satisfying Alarcon Segovia criteria.

KEYWORDS

MCTD(Multiple Connective Tissue Disease), Pulmonary Hypertension, Sclerodactyly, Raynaud's Phenomenon, U1RNP antibodies

INTRODUCTION

MCTD is a rare entity which embraces features of SLE, scleroderma, polymyositis , dermatomyositis with an incidence rate that varies between 0.2 to 1.9 in 100000 patients per year with male to female ratio of 1:5 . There are no distinct unique features to MCTD. It can present with any of the overlapping features of SLE , scleroderma, Polymyositis , Dermatomyositis and/or Rheumatoid Arthritis which can appear at any time throughout the course of the disease. Presence of anti RNP antibodies is a requisite criterion with most common clinical associations are Raynaud's phenomenon, sclerodactyly, hand edema, arthralgia .

Figure 1: Tightening Of Skin Of Face With Restricted Mouth Opening

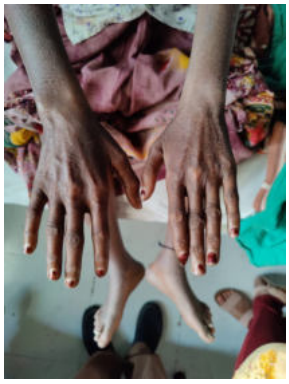

Figure2: Tightening Of Skin With Ulcerative Lesions Over Interphalangeal Joint Skin,sclerodactyly

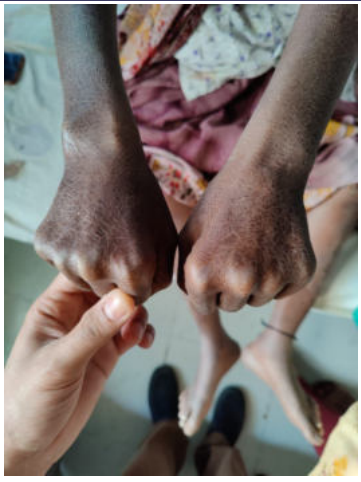

Figure 3: Hyperpigmentation Of Skin Of Upper Limb

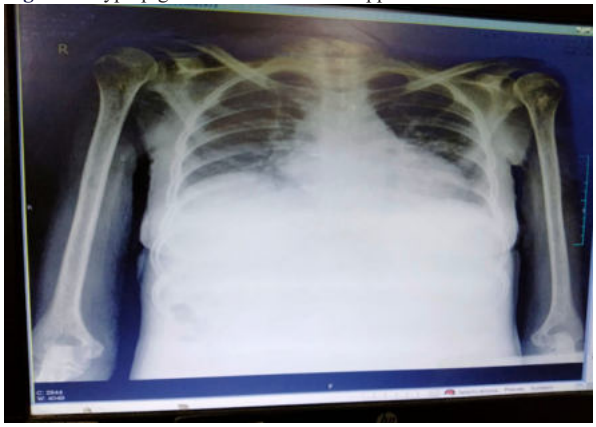

Figure 4: Chest X Ray With Prominent Upper Lobe Pulmonary Vessels S/o Pulmonary Hypertension And Cardiomegaly.

Case Study:-

We present a 38 year old female brought by relatives to our tertiary care hospital with chief complaints of tightness, dryness, hyperpigmentation of skin, tightness of mouth, tingling numbness and pain of fingers while handling cold water since 6 months, breathlessness on exertion since 1 month . She had history of swelling

of hands 6 months back for which she was treated outside conservatively. Patient had no history of cough, cold , fever , chest pain , palpitations.No history of any drug reaction, allergy . No past surgical or medical history . No comorbidities . Patient was completely immunized .Patient consumed mixed diet and had no addictions. On admission patient was conscious, oriented, afebrile , averagely built , lying comfortably in bed. Vitally Blood pressure- 120/80 mmHg , pulse rate – 88/min regular , normal volume, no radio radial or radio femoral delay with all peripheral pulsations felt . Oxygen saturation – 98% on room air . Skin over face, upper limb and lower limb was tightened and hyperpigmented with restricted mouth opening. Finger examination showed sclerodactyly and ulceration over skin of interphalangeal joints of both hands without any deformity.

Systemic Examination:
Cardiovascular examination – Loud P2 in pulmonary area with grade lll pansystolic murmur in tricuspid area .

Respiratory system examination: Air entry was equal on both sides with normal breath sounds heard over right and left lung fields with no adventitious sounds.

Neurological examination: Patient was conscious oriented to time, place and person with no neurological deficits.

Per abdomen examination : No organomegaly . No tenderness guarding or rigidity present on palpation. Bowel sounds were normal.

Investigations-
Haemoglobin – 10.4 , MCV – 96 , WBC count – 7400, ESR – 16, ANA- 5.5 (positive),

U1RNP – 93 (strong positive, titre >1:1600) , CRP – 4.1 , C3 – 227 (84-168) , C4 – 9.81(9-36)

S. Sodium- 137, S. potassium – 3.7, urea – 13, S creatinine- 0.6, Total bilirubin- 0.4 , Direct bilirubin - 0.2 , Indirect bilirubin -0.2, SGOT – 21, SGPT – 14 , Total protein- 7.1 , albumin- 3.9, globulin 3.2 , INR – 0.9, HIV – non reactive, HbsAg – non reactive, HCV – non reactive. ECG - HR- 96/min, Left axis deviation , no significant ST T changes , chest x ray - prominent upper lobe pulmonary vessels suggestive of Pulmonary Hypertension. Evidence of cardiomegaly.

2D Echo- normal RV , LV systolic function, severe pulmonary Hypertension, severe Tricuspid regurgitation. RA factor- negative , anti Sm antibodies- negative, anti SSa antibodies – negative,
Anti SSb antibodies- negative, anti Scl 70 – negative, xray both hands PA – wnl.

Based on clinical and laboratory findings, patient was diagnosed as a case of MCTD as per Alarcon Segovia Criteria. Patient was started on corticosteroids , phosphodiesterase -5 inhibitors (sildenafil, bosentan) for pulmonary Hypertension and other symptomatic treatment.

DISCUSSION-
Clinical features of MCTD often develop over several years and complete clinical findings are rarely present at the start of the disease. The most common manifestations are Raynaud's Phenomenon, Sclerodactyly, Swelling of hands , arthralgia . The most sensitive classification criterias for clinical diagnosis of MCTD are Alarcon segovia and Kasukawa criteria.

Alarcon Segovia Criteria:
Serology(must be present) : Anti RNP titre more than or equal to 1:1600
Clinical criteria:
more than or equal to 3 clinical criteria
1 Swollen hands
2 Synovitis
3 Myositis
4 Sclerodactyly
5 Raynaud's phenomenon

Diagnostic criteria may help define patient with MCTD but patient may not fulfill the diagnostic criteria at their initial presentation as patient may develop new clinical and laboratory features during the course of the disease as noted in the observation. Given that clinical features of MCTD tend to occur sequentially over years , long term follow up of patients is important. In such patients corticosteroids are beneficial for milder manifestations . Raynaud's phenomenon can be treated conservatively by keeping extremities , head , and other body parts shielded from exposure to cold . In severe or refractory cases, vasodilators including calcium channel blockers , phosphodiesterase – 5 inhibitors (such as sildenafil) can be employed to control symptoms and ischemic complications. Immunosuppressive and cytotoxic therapies are reserved for most severe cases of internal organ involvement. Pulmonary involvement and severe complications like Pulmonary Hypertension are significant morbidities that can begin asymptomatically and if not detected timely and not treated promptly can affect the prognosis of the disease leading to worse outcomes.

CONCLUSIONS :-
MCTD can present as a distinct entity with overlapping features which might not be present concurrently . All the patients with MCTD should be followed closely and managed according to their clinical and laboratory assessments and specific underlying internal organ involvement. Moreover clinical features of MCTD can change over time thus reappraisal of management strategies is needed at each patient encounter. Timely diagnosis of MCTD with clinical and laboratory findings ,long term and close follow up of patients for early detection of complications and effective treatment is important to prevent morbidity and mortality.

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