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Case Report

Paediatrics Section

MDA5 Juvenile Dermatomyositis Misdiagnosed as Childhood Psoriasis: A Case Report

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ABSTRACT

Juvenile Dermatomyositis (JDM) is a rare autoimmune disorder which mainly affects the skin and skeletal muscles. Characteristic features of this rare disorder include Gottron papules, heliotrope rashes and proximal muscle weakness. MDA5 positive JDM is usually associated with interstitial lung disease in later part of life. The authors report a case of an 11-year-old male child who presented to the Paediatric Department with complaints of rashes over extensor surfaces, Gottron papules and pain and weakness in all limbs. The child was on treatment from the Dermatology Department as a case of psoriasis for last six months. During hospital stay after detailed work-up, the child was diagnosed to have JDM. Anti-melanoma Differentiation-Associated gene 5 (Anti-MDA5) antibodies were positive in the present case. The child was treated with immunosuppressive drugs with supportive care and is currently on treatment for the last one and a half years and is largely asymptomatic. JDM requires meticulous clinical examination and specific investigations so that timely diagnosis can be made to ensure appropriate treatment. In the present index case, the authors describe the typical clinical presentation, newer investigative modalities like myositis specific antibody and treatment outcomes of this rare condition.

Keywords: Gottron papule, Inflammatory myositis, Melanoma differentiation-associated gene 5 antibody

CASE REPORT

An 11-year-old male child presented to the Paediatrics Outpatient Department (OPD) with complaints of easy fatiguability, rashes over extensor surfaces since one year and pain and weakness in all limbs since last two months. Initially, he was managed as psoriasis at the Dermatology OPD and was on treatment for last six months where he didn't show any improvement.

Rashes were maculopapular, itchy, started on upper extremities (arms, hand and on knuckles) and progressed to involve the thigh. Limb pain was gradual in onset, dull aching in nature, moderate in intensity, aggravated by physical activity and this rendered the child unable to attend school for last two months. Weakness was gradual in onset, progressive and symmetrical in nature, involving the proximal aspect of upper and lower limbs. There was no history of any hospitalisation for any cause. He was immunised for age and belonged to lower socioeconomic strata as per modified Kuppuswamy scale [1]. There was no family history of any other chronic illness. On physical examination, he was afebrile (Temperature-98.5°F) with heart rate-92/min, respiratory rate-24/min, blood pressure-110/72 mmHg. Local examination showed erythematous maculopapular rash on photoexposed areas, erythematous papules over metacarpophalangeal and interphalangeal joints (Gottron papules) [Table/Fig-1], lymphadenopathy in cervical and submandibular areas (multiple lymph nodes measuring around 10 mm, non tender). There were inflamed and swollen areas present around fingertips with tenderness in all limbs (muscle tenderness). Systemic examination was normal for all systems.

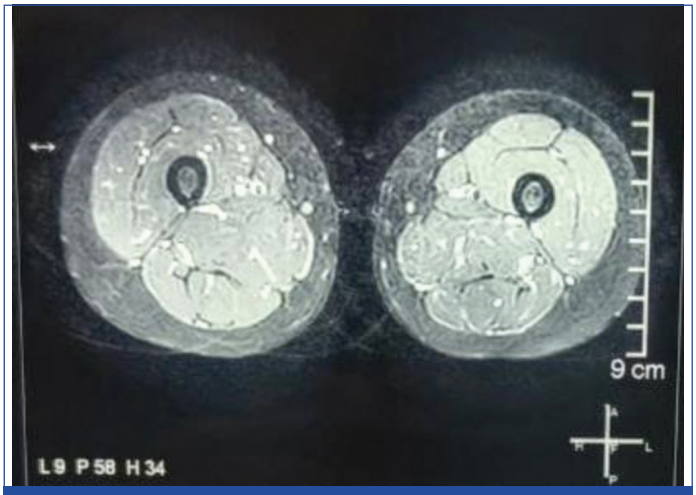
[Table/Fig-1]: Small sized papules at metacarpophalengel joint.

Investigation revealed raised Erythrocyte Sedimentation Rate (ESR), Lactate Dehydrogenase (LDH), Creatine Kinase (CK) with positive anti MDA-5 antibody [Table/Fig-2]. Magnetic Resonance Imaging (MRI) of the thigh was normal [Table/Fig-3], but High Resolution Computed Tomography (HRCT) of the thorax showed areas of consolidation with ground glass opacities and tractional bronchiectasis [Table/Fig-4]. Juvenile Dermatomyositis (JDM), Idiopathic inflammatory myopathies, non inflammatory myopathies, skin conditions like psoriasis and eczema were considered. Definitive diagnosis of JDM was made on the basis of Consensus Group of European League against Rheumatism/ American College of Rheumatology (EULAR/ACR) 2017 criteria [2]. Points in favour are- muscle pain and tenderness, easy fatigability, skin lesion (Gottron papules, scaly and dry rough skin, inflamed and swollen area around fingertips), elevated ESR, CK, LDH and positive myositis profile (Anti MDA5).

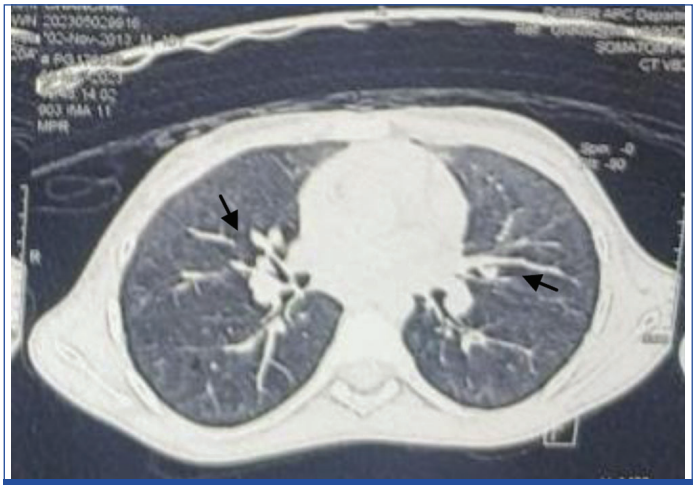
The patient initially was treated with intravenous methylprednisolone pulse therapy (30 mg/kg/day) for three days and then oral prednisolone at 1 mg/kg/day was started. Prednisolone was continued at the same dose for two month and then was tapered gradually over next two months and subsequently stopped. Along with the steroids, methotrexate injection was given subcutaneously 15 mg/m^2 weekly and has been continued since then and the treatment plan includes giving it for a minimum of two years. The patient was also advised oral calcium and vitamin D supplementation and was advised to avoid direct sunlight exposure along with use of sunscreen. Follow-up visits have shown weight gain of 5 kg, improvement in skin lesions and with the help of physiotherapy muscle weakness has also improved allowing the child to resume school. Physiotherapy in the form of walking, passive stretching and range of motion exercises were done three times per week for eight weeks followed by two times per week for eight weeks. Parents were advised to continue the exercises at home. Currently, the child is on subcutaneous methotrexate injection weekly and is doing well. Parents' consent was taken for publication purposes (as the patient is minor).

Laboratory investigation	Pre-treatment	Post-treatment
Complete Blood Count (CBC)	Hb-12.2 g/dL WBC-6880/cu mm Platelet count-1.93 lacs	Hb-12.6 g/dL WBC-5800/cu mm Platelet count-2.56 lacs
Blood urea Serum creatinine	35 mg/dL 0.7 mg/dL	24 mg/dL 0.6 mg/dL
Erythrocyte Sedimentation Rate (ESR)	39 mm in 1st hour	20 mm in 1st hour
Lactate Dehydrogenase (LDH)	408 U/L	112 U/L
Creatine Kinase (CK)	105 U/L (normal range-20-30 U/L)	30 U/L
HIV serology and serum IGA TTG	Negative	
Autoimmune work-up	ANA- negative ANCA (P-ANCA and C-ANCA)-negative	
Myositis profile	Anti-MDA5 antibody positive	-
Imaging		
USG neck	Multiple hypoechoic lymph nodes in bilateral cervical region (left > right), largest one 13*4 mm.	
MRI thigh	Normal	
HRCT thorax	Areas of consolidation with ground glass opacities noted in bilateral lung parenchyma mainly in subpleural location with areas of septal thickening and tractional bronchiectasis	

[Table/Fig-2]: Investigations supporting the diagnosis of Juvenile Dermatomyositis (JDM).
ANA: Antinuclear antibody; ANCA: Antineutrophilic cytoplasmic antibody



[Table/Fig-3]: MRI of the thigh revealed no significant abnormality.



[Table/Fig-4]: HRCT of the chest done showed areas of consolidation with ground glass opacities noted in bilateral lung parenchyma mainly in subpleural location with areas of septal thickening and tractional bronchiectasis.

DISCUSSION

The JDM is the most common inflammatory myositis in children with an incidence of 2.5 cases per million children per year [3]. Peak age of onset of disease is 5-10 years with male to female ratio of 1:2 [4]. The aetiology is idiopathic; however, environmental factors and immune dysfunction are believed to have a role in its aetiopathogenesis. It mainly affects the skin and skeletal muscles. Typical clinical presentation includes characteristic skin lesions (Gottron papules, heliotrope rash) with proximal muscle weakness. However, the index case presented with muscle pain and tenderness, muscle weakness, skin lesions (Gottron papules, scaly and dry rough skin, inflamed and swollen area around fingertips). Mussa F et al., reported a case of an eight-year-old girl child who presented with typical features of JDM and has a rare association with autoimmune thyroiditis. The present case implicates the characteristic manifestation of JDM can be clearly misdiagnosed and thus, under-reported [5]. Yeung T-W et al., reported a case of a 16-year-old female who had typical features of JDM with anti MDA5 antibody was positive [6].

Initially, diagnostic criteria was based on Bohan and Peter classification given in 1975 which included: classic rash, proximal muscle weakness, elevated muscle enzymes, electromyographic changes and muscle biopsy [7,8]. Latest diagnostic criteria given by EULAR/ACR in 2017 which includes skin rash, symmetrical proximal muscle weakness, elevated skeletal muscle enzymes, specific Electromyography (EMG) changes, muscle biopsy, myositis specific antibodies and MRI evidence of myositis [2]. The index case had elevated ESR, CK, and LDH. Myositis specific antibodies test done was positive (Anti MDA5 positive). Myositis Specific Autoantibodies (MSAs) have been detected in up to 54% of children with JDM in the United States [9]. The Anti-melanoma Differentiation Associated gene-5 antibody (Anti MDA5) is associated with interstitial lung disease.

Based on EULAR/ACR criteria, a definitive diagnosis of JDM was made. The management of JDM according to previously published literature includes steroids and antimetabolites like methotrexate [10,11]. The index case was managed with methylprednisolone pulse therapy followed by oral prednisolone, subcutaneous methotrexate injection with calcium and vitamin D supplementation. Currently, the child is on regular follow-up and doing well. JDM is a rare presentation and required identification of characteristic lesion (Gottron papule) and appropriate and timely management to improve outcomes.

CONCLUSION(S)

To conclude, the child was misdiagnosed as a dermatological case based on clinical presentation of rashes and systemic complaints were overlooked. Subsequent to unresponsiveness to treatment, the child was presented to the Paediatric Department where elaborate clinical examination and targeted investigations led to diagnosis and appropriate management. Myositis specific antibodies (Anti MDA5) played a pivotal role in making the diagnosis and can be considered an important tool to diagnose this rare autoimmune disorder.

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