Case Report

Juvenile dermatomyositis in a 6-year-old girl: A case report

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ABSTRACT

Juvenile dermatomyositis (JDM) is the most common inflammatory myositis in children, distinguished by proximal muscle weakness and a characteristic rash. This 6-year-old child presented to the dermatology clinic with a heliotropic rash and later on weakness worsened making her non-ambulant. Genetically pre-disposed children with susceptibility to JDM have upregulation of gene products controlled by type I interferon. Hence, the etiology of JDM is multifactorial, such as genetics and environmental triggers. The diagnostic criteria comprise of heliotropic rash, symmetrical proximal muscle weakness, elevated biomarkers, electromyography, and histopathological changes in muscle. The child was started on intravenous immunoglobulin and immediate clinical remission was attained which emphasized the role of early aggressive institution of therapy in JDM.

Key words: Juvenile dermatomyositis, Proximal myopathy, Heliotrope rash, Gottron papule

Juvenile dermatomyositis (JDM) is the most common childhood inflammatory myopathy which is a systemic capillary vasculopathy. Patients present with proximal muscle weakness, raised muscle enzymes, and pathognomic skin rashes such as heliotrope rash and Gottron's papules. Complement-mediated damage of vessels is a major mechanism contributing to the pathology. Incidence is 2–3/million/year and from 1995 to 1998 in the US ranged from 2.5 to 4.1 cases/million children which makes it a much rarer yet challenging situation to tackle [1,2].

This motivated us to report our case as literature was lacking from South India. Our case had classic clinical features and showed a drastic response to pulse methylprednisolone and intravenous immunoglobulin (IVIG). A detailed study regarding JDM-associated autoantibodies by Vignesh *et al*. [2] in 2022 and long-term outcomes in children with JDM by Sharma *et al*. [3] shows us how rare the disease is and how it mandates further studies.

CASE REPORT

A worried mother to 6-year-old twins came to us with the girl child having generalized rash and edema of 1-month duration. The girl was treated at a dermatology clinic for rash and at a homeopathic clinic for fatigue with no improvement in her symptoms. She came to us with a characteristic itchy erythematous photosensitive heliotropic rash around both eyes which became violaceous over time (Fig. 1) and progressed to elbows, knees, neck, and chest, sparing the abdomen and genital area. She had generalized edema and weakness which limited her mobility. She was the second dichorionic diamniotic (DCDA) twin with a twin brother and elder sister all immunized up to age. Edema was insidious in onset along with rash which was followed by myalgia and weakness which predominantly affected the proximal muscles of both upper and lower extremities, and pain, swelling, and papular eruptions over bilateral knee joints for 1 month. Her past medical and family history was unremarkable.

On examination, she had violaceous rash around the eyes and cheeks (heliotrope rash) and erythematous patches were noted on the metacarpophalangeal and interphalangeal joints of the dorsum of both hands (Gottron's papules Fig. 2) and "shawl sign" over chest wall (Fig. 3a). There was swelling and tenderness of small joints of the hand and feet with a mildly restricted range of motion. She had symmetrical proximal muscle weakness in both the lower limbs with hypotonia which caused difficulty in combing hair getting up from her bed, and difficulty in walking. Power at the hip joint was 2/5, at the knee 3/5, and 4/5 in all upper limb joints on both sides. All deep tendon jerks were not elicitable with otherwise normal neurological examination. The oral cavity showed extensive mucosal candidiasis (Fig. 3b) and a congested posterior pharyngeal wall. Respiratory examination revealed fine inspiratory crepitation over the lower lobes on both sides. She also had erythematous scaly plaques over the elbow

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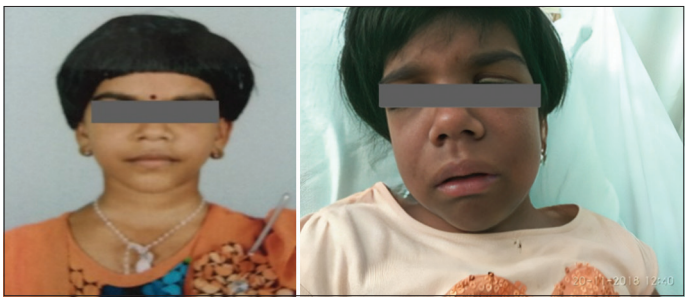

Figure 1: Clinical photograph of child before and after the onset of rash

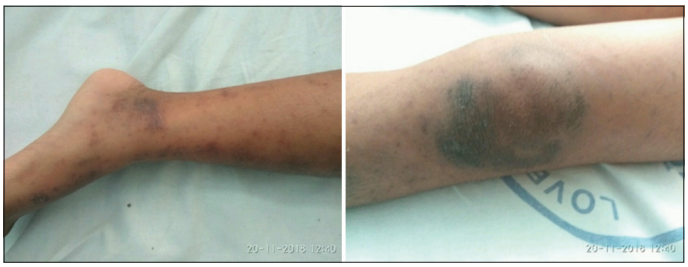

Figure 2: Gottron's papules

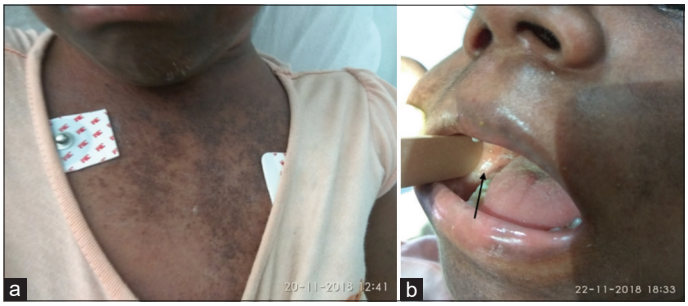

Figure 3: (a) shawl sign; (b) oral candidiasis

and the knee joints and hypertrichosis and hyperpigmentation over the forehead, neck, and cheeks. She also had multiple tender nodules 0.5 cm×0.5 cm on elbows and knees bilaterally.

Initial investigations revealed normocytic normochromic anemia (Hb=9.7 g/dL,) with a white cell count of 7800/μL and a negative C-reactive protein with erythrocyte sedimentation rate of 11 mm/h. Liver function tests showed mild elevation of transaminases (serum glutamic-oxaloacetic transaminase [SGOT]=148 U/L; serum glutamic pyruvic transaminase=41 U/L) with elevated lactate dehydrogenase (LDH)=1877 (140–280 U/L). Renal function, electrolytes, and urinalysis were normal. Creatine phosphokinase (CPK) values were very high (2560 U/L [55–170 U/L]) which is pathognomonic of muscle involvement. Autoantibodies were tested which came back negative.

She was started on 2 g/kg of immunoglobulin and prednisolone while waiting for electromyography (EMG). Drastic symptomatic improvement was noted as myalgia subsided and she became playful and started sitting on her bed (Fig. 4). Biochemical markers improved like CPK, which was 2560 U/L which gradually lowered to 2160 U/L then to 1159 U/L and in 1 week to 930. LDH which was initially 1877 U/L lowered to 1617 U/L in 1 week and SGOT remained high at 146 even at the end of 1 week.

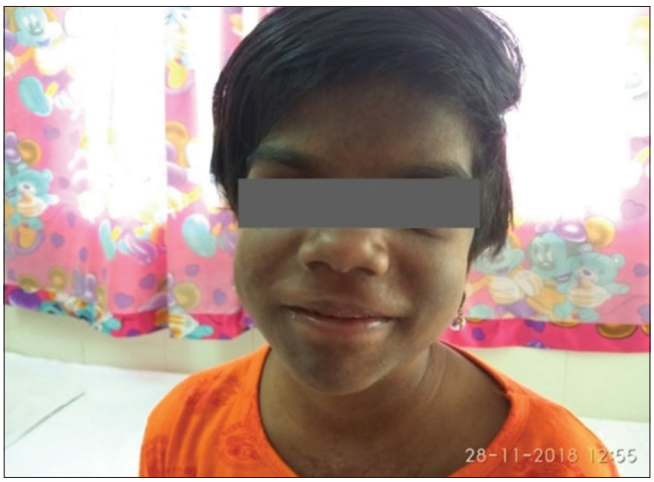

Figure 4: After intravenous immunoglobulin treatment

DISCUSSION

JDM is the most common inflammatory myositis in children [4]. For children, 2–17 years of age, the estimated annual incidence rates from 1995 to 1998 in the US ranged from 2.5 to 4.1 JDM cases per million children, and the 4-year average annual rate was 3.2/million children (95% confidence interval 2.9–3.4) [5]. Inflammatory cell infiltrates resulting in vascular inflammation and leakage are the underlying pathology in this disorder.

The etiology of JDM is multifactorial, based on genetic predisposition and environmental triggers. Although classified under inflammatory myopathy some cases lack muscle weakness despite skin lesions characteristic of JDM and this is called juvenile clinically amyopathic dermatomyositis (JCADM). JCADM comprises juvenile amyopathic dermatomyositis and juvenile hypomyopathic dermatomyositis [6]. Rheumatic diseases of childhood exhibit extreme photosensitivity to ultraviolet light exposure with generalized erythema in sun-exposed areas. Such erythematous lesions seen over the chest and neck are called as the "shawl sign." Erythema is also commonly seen over the knees and elbows. The characteristic heliotrope rash is a blue-violet discoloration of the eyelids that may be associated with periorbital edema.

The common history given by parents is of infection in the 3 months before the diseases, such as upper respiratory and gastrointestinal symptoms. Children present with clinical features of muscle weakness in 90–100% of the time and others include dysphagia or dysphonia, muscle atrophy, muscle pain, and tenderness. Skin lesions include Heliotrope rash of eyelids, Gottron's papules, erythematous rash of malar/facial area, periungual capillary changes, photosensitive rash, ulcerations, calcinosis, lipodystrophy, Raynaud phenomenon arthritis and arthralgia, joint contractures and fever. Fatigue is a common presenting complaint in JDM [4]. While signs suggestive of a rheumatic etiology, such as heliotropic rash, and Gottron's papules are specific to JDM the symptoms are non-specific, such as arthralgia, weakness, and fatigue, which makes the patient delay in attaining medical care. Lack of a specific autoantibody also makes it more of a clinical diagnosis although biochemical

markers, such as complete blood count, CPK, alanine transaminase, aspartate transaminase, LDH, aldolase, antinuclear antibodies, and gag reflex aid us in evaluating when we suspect JDM [7].

Muscle-derived enzymes (creatine kinase, aldolase, aspartate aminotransferase, alanine aminotransferase, and LDH) are elevated and reflect muscle inflammation. A more specific and initial peak of alanine aminotransferase followed by creatine kinase peak is seen. Although imaging studies are not included in diagnostic criteria T2 magnetic resonance imaging helps to identify active sites of disease. EMG shows signs of myopathy (increased insertional activity, fibrillations, and sharp waves) as well as muscle fiber necrosis (decreased action potential amplitude and duration) [1]. Muscle biopsy shows focal necrosis and phagocytosis of muscle fibers, fiber regeneration, endomysial proliferation, inflammatory cell infiltrates and vasculitis, and tubuloreticular inclusion bodies within endothelial cells [4].

Diagnosis of JDM is based on clinical biochemical, EMG, and muscle biopsy findings which are solely based on criteria developed by Peter and Bohan in 1975 although 2017 EULAR/ACR criteria addresses inflammatory myopathies in the absence of muscle biopsy [8,9]. Bohan and Peter's criteria include a classic heliotrope rash of the eyelids, Gottron's papules Plus 3 of the following i.e., weakness which is symmetric and proximal, muscle enzyme elevation, such as creatine kinase, aspartate aminotransferase, LDH, aldolase and electromyographic changes and fourth being muscle biopsy necrosis or inflammation although biopsy is rarely done due to its invasiveness [9]. Worldwide, Peter and Bohan still hold better acceptance [10] and with some modifications are still being used widely (Table 1).

Management of JDM should always be aggressive to limit long-term disabilities. Corticosteroids are still the mainstay of treatment. In a clinically stable child without debilitating weakness, oral prednisone at 2 mg/kg/day (maximum 60 mg daily) is usually started. Other treatment modalities include methotrexate (MTX), IVIG, cyclosporine, and mycophenolate mofetil. Corticosteroids have altered the course of the disease, lowering morbidity and mortality. MTX decreases the length of treatment with corticosteroids, thereby reducing morbidity from steroid toxicity. Intravenous gamma globulin is frequently used as an adjunct for the treatment of severe disease. MTX is commonly used in the treatment of JDM as a steroid-sparing agent, with efficacy in 70% of patients [4]. Childhood Arthritis and Rheumatology Research Alliance has been a single access point for clinicians in standardized treatment approaches. Initial therapy comprises a combination of steroids and MTX, with an escalation of therapy for resistant disease [11].

Long-term outcomes in childhood myositis from a patient perspective by Boros *et al*. found high self-perceived reported rates of ongoing muscle disease, skin disease, arthritis, and the need for immunosuppressive medication use in long-term follow-up and it highlights opinions and needs of adolescents and young adults [12].

The latest advances in JDM treatment are the use of biological agents, such as tumor necrosis factor inhibitors, rituximab, abatacept, and tocilizumab. Rituximab is a monoclonal antibody directed against CD20, a surface marker for B cells. Abatacept is a genetically engineered fusion protein. Janus kinase inhibitors, such as tofacitinib, baricitinib, and ruxolitinib. Lenabasum is an oral synthetic cannabinoid receptor type 2 agonist for pruritis under consideration. The treatment of cutaneous disease in JDM also includes avoidance of ultraviolet B exposure [13].

Table 1: The Bohan and Peter criteria for DM and PM

First, rule out all other forms of myopathies
1. Symmetrical weakness, usually progressive, of the limb-girdle muscles with or without dysphagia and respiratory muscle weakness
2. Muscle biopsy evidence of myositis
Necrosis of type I and type II muscle fibers; phagocytosis, degeneration, and regeneration of myofibers with variation in myofiber size; endomysial, perimysial, perivascular, or interstitial mononuclear cells
3. Elevation of serum levels of muscle-associated enzymes (CK, LDH, transaminases, aldolase)
4. EMG triad of myopathy
a. Short, small, low-amplitude polyphasic motor unit potentials
b. Fibrillation potentials, even at rest
c. Bizarre, high-frequency repetitive discharges
5. Characteristic rashes of dermatomyositis

PM: Polymyositis, DM: Dermatomyositis, LDH: Lactate dehydrogenase, CM: Creatine kinase, EMG: electromyography. Definite PM: all first four elements, probable PM: 3 of first 4, possible PM: 2 of first 4. Definite DM: rash *plus* 3 others, probable DM: rash *plus* 2 others, possible DM: rash *plus* 1 other

CONCLUSION

We have presented the case of a DCDA twin girl with classical clinical findings of JDM. It is important to diagnose and initiate immunosuppressive treatment early in the natural history of the disease to reduce the burden of long-term complications. This case has been reported for its rarity and to emphasize the importance of early and aggressive treatment to prevent long-term disease sequelae.

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