Juvenile Dermatomyositis in an Indian girl: a rare condition in childhood - 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. Inflammatory cell infiltrates result in vascular inflammation, the underlying pathology in this disorder. Its incidence is approximately 3 cases/1 million children/year. Peak age of onset is between 4 and 10 years. Etiology is multifactorial, based on genetic predisposition and an unknown environmental trigger. The cutaneous manifestations are the most important aspect of this disease, and their correct evaluation is important for early diagnosis. A 7 year old female child with proximal muscle weakness of all four limbs, heliotrope rash of the eyelids and Gottron papules was diagnosed to have JDM. This case report aims to highlight that it is a rare but potentially life threatening autoimmune disease of childhood. So it has to be diagnosed early and treatment should be initiated to prevent long term complications. Children with this condition appear able to repair inflammatory damage to vasculature and muscle.
Introduction

Juvenile dermatomyositis (JDM) is a rare but complex immune mediated disease of childhood, primarily affecting proximal muscles and skin.\(^1\) Although the cause remains unknown, it is clear that genetic and environmental influences play a role in the etiology.\(^1\)

It is characterized by profound muscle weakness in addition to skin lesions, calcinosis, and underlying vasculopathy.\(^2\)

Calcinosis is a major cause of morbidity in this condition and has previously been linked to younger age at disease onset and more persistent disease activity.\(^3\) It is a soft tissue calcification seen in more severe cases and its prevalence ranges between 40-60%.\(^4\)

It is part of a heterogeneous group of muscle diseases called idiopathic inflammatory myopathies.\(^5\)

Correct evaluation of cutaneous manifestations and myositis-associated auto antibodies should help the clinician in early diagnosis, for a quick recognition of cutaneous signs that may be the symptom of onset before muscle inflammation.\(^6\)

Whole body Magnetic Resonance Imaging (WB-MRI) provides additional information to clinical evaluation and represents a promising tool to estimate total inflammatory burden, tailor treatment and monitor its efficacy.\(^7\)

For typical cases of JDM, regardless of severity, almost all respondents used Corticosteroids and another medication, Methotrexate (MTX) being the most commonly used.\(^8\)

Outcomes have improved with advances in medical treatment; the mortality rate has decreased to \(<2\%\), but 60–90\% of patients develop disease damage after 7–17 years of follow-up.\(^10\)

Case Description

A seven year old female child, admitted to Vani Vilas hospital of Bangalore Medical College and Research Institute, first born to a non-consanguineous parentage belonging to lower middle socioeconomic status according to modified Kuppuswamy scale, with uneventful antenatal, natal and neonatal period and normal development, immunized upto date as per the National Immunization schedule of India, presented with proximal muscle weakness of all four limbs since one and a half months, pain and swelling in both the lower limbs since one month and photosensitive rash over the face and upper limbs since one month. There was also history of altered speech in the form of nasal speech and difficulty in swallowing both solids and liquids.

On examination, thinness was present (Body Mass Index 13.4). Child had periorbital hyperpigmented rash with periorbital edema (Heliotrope rash), Gottron’s papules over bilateral proximal interphalangeal joints of hands. Power of proximal muscles (2/5) was less compared to distal muscles (3/5). In addition to this, child had a waddling gait.

Investigations included a complete haemogram, renal function tests, urine routine which were normal. Liver function tests showed an elevated Aspartate transaminase 397 U/L and Alanine transaminase 175 U/L. CRP was negative, ESR-50, ANA profile negative.

Additional investigations included: Creatine phosphokinase > 4267 U/L, Lactate Dehydrogenase levels 1504 U/L, MRI muscle which was normal. However muscle biopsy showed rounding and variation in fibre size, myophagocytosis, myonecrosis, fibres with internalized nuclei and atrophic rounded fibres, perifascicular atrophy prominent, dense endomysial, interstitial and perivascular inflammation in endo, peri and epimysial vessels of lymphocytes, plasma cells and macrophages. These findings clinched the diagnosis of Dermatomyositis.
Electroneurogram showed features suggestive of motor axonal neuropathy of right median, ulnar and common peroneal nerves.

Differential diagnosis considered was Collagen vascular disease like Systemic Lupus Erythematosus (SLE), but SLE was ruled out based on American College of Rheumatology 1997 revised criteria. ANA profile was also negative.

Diagnosis of Dermatomyositis was confirmed since the diagnostic criteria included classic rash: Heliotrope rash of the eyelids, Gottron papules, plus 3 of the following:

1) Symmetric and proximal muscle weakness
2) >= 1 muscle enzyme elevation: Creatine kinase, Aspartate transaminase, Lactate dehydrogenase.
3) Muscle biopsy: showed necrosis, inflammation

Child was given high dose pulse Intravenous Methylprednisolone (30 mg/kg/day for 3 days), then followed up with oral Prednisolone at 1 mg/kg/day. Weekly oral Methotrexate (0.5-1 mg/kg) was also used as a steroid sparing agent. Folic acid was typically given with Methotrexate, started at a dose of 1 mg daily to reduce toxicity and side effects of folate inhibition.

At present on follow up, muscle weakness of the child has reduced.

Discussion

In contrast to adults with dermatomyositis, children with JDM are more likely to have complications that are thought to indicate a vasculopathic process, such as severe skin disease, with ulceration or calcinosis, gut vasculopathy or central nervous system disease.\(^1\)

JDM is an autoimmune connective tissue disease occurring in children less than 16 years old.\(^5\) The index sign of heliotrope rash is often difficult to visualise in the black skin.\(^5\)

Intravenous immunoglobulin (IVIG) was used more frequently for more severe disease, for refractory disease, and for prominent cutaneous disease.\(^8\) Hydroxychloroquine was often used in milder cases and those principally characterized by rash.\(^8\)

Steroids have to be slowly tapered over a period of 12-24 months, after indicators of inflammation (muscle enzymes) normalize and strength improves. Children receiving prolonged steroid therapy are prone to complications such as cessation of linear growth, weight gain, hirsutism, adrenal suppression, immunosuppression, striae, cushingoid fat deposition, mood changes, osteoporosis, cataracts, avascular necrosis, and steroid myopathy.\(^9\)

Most complications from this disease are related to prolonged and severe weakness, including muscle atrophy, to cutaneous calcifications and scarring or atrophy, and to lipodystrophy.\(^9\) Children with acute and severe weakness are at risk for aspiration pneumonia and respiratory failure.\(^9\) Crampy abdominal pain and occult GI bleeding may indicate bowel wall vasculitis and lead to ischemia, GI bleeding and perforation.\(^9\) Cardiac involvement is rare but includes arrhythmias.\(^9\)

The mortality rate has decreased since the advent of Corticosteroids, from 33% to currently about 1%. At 7 years of follow up, 75% of patients have little to no residual disability, but 25% continue to have chronic weakness and 40% have chronic rash.\(^9\)

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Seven year old female with Juvenile Dermatomyositis

Heliotrope rash of the eyelids with periorbital edema

Gottron's papules-shiny plaques over proximal interphalangeal joints of hands

Reduced periorbital puffiness after 2 weeks of steroid therapy