

Dermatomyositis a Diagnostic Delimma: An Interesting Case Series and Review of Literature

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Abstract

Dermatomyositis is a rare idiopathic inflammatory myopathy with characteristic cutaneous findings that occur in children and adults. We report a case series of dermatomyositis rare disease and the dilemma associated with the diagnosis. All the three cases we discussed were difficult to diagnose and had diagnostic dilemma. This case series highlights the investigation required to diagnose such disease. The main aim of the article is to create awareness among the orthopaedicians to have clinical suspicion of dermatomyositis in cases of diffuse dystrophic calcification.

Keywords: Dermatomyositis; Heliotropic rash; Muscle weakness; Stiffness; Steroids

Introduction

Dermatomyositis is the least common among idiopathic inflammatory myopathy with characteristic cutaneous findings that occur in children and adults. The condition can affect both adults and children. In adults, dermatomyositis usually occurs from the late 40s to early 60s. In children, it most often appears between 5 and 15 years of age. Dermatomyositis affects more females than male. It affects skin, muscle, subcutaneous tissue & blood vessels. Muscle weakness may occur concurrently or after weeks to years [1]. DM may be associated with systemic manifestations like malaise, arthralgia, dyspnoea, etc. However, subcutaneous calcifications are especially common in children [2]. The outcome of this disease is altered by early diagnosis and aggressive pharmacologic corticosteroid treatment [3]. It is a rare diagnosis which is commonly missed, leading to delay in diagnosis. The delayed diagnosis can leave the case untreated and therefore is often accompanied by continued and increasing inflammation with potential systemic damage [4]. The symptoms can vary from malaise, skin lesion to muscle weakness and wasting. Later on systemic involvement can be seen. The skin biopsy and muscle biopsy was done by dermatologist.

Case 1

An 8 year male child reported in Orthopaedics Opd with complains of stiffness in all joints and generalized weakness. The child developed generalized weakness 3years back, which was insidious in onset and progressive in nature. Initially child had weakness in lower limb, which leads to difficulty in walking, getting up from squatting position and climbing stairs. The muscle weakness then progressed and involves upper limbs. The child had difficulty in overhead abduction of both upper limbs. The proximal muscle weakness progressed and child was bed ridden. For the past 6 months child was walking with support for 20-50 meters only. The weakness was lower motor neuron type of

weakness associated with muscle wasting. There was no sensory loss associated with it. Bladder and bowel of the child was never involved. The child also skin lesion for 3 years. Skin lesions include darkening on his face, eyelids & cheeks. Later child developed hyperpigmentation and thickening of skin around knuckles, wrist, elbow and knees and bilateral dorsum of feet. Skin became scaly, shiny and edematous with loss of hair. On examination, multiple subcutaneous nodules, heliotropic purple discoloration of upper eyelids, Gottron's sign, rash on extensor surface of elbow and mechanics hand present. The child developed stiffness of all joints mainly bilateral knee, hip, ankle and elbow joints (Figure 1). On the basis of all these clinical findings myositis ossificans progressiva and dystrophic calcinosis were thought of. Investigations were done to ascertain the diagnosis. CBC showed TLC of 15400/mm³, Hb of 7.6 g/dl, peripheral smear suggested of microcytic hypochromic anaemia. Other laboratory investigations showed LDH of 470 U/L, CPK of 600 U/L, CK-MB of 96 U/L, iCalcium of 1.07, ALP of 560 U/L and ANA was positive. EMG showed increased spontaneous muscle activity with fibrillations, complex repetitive discharges. X-rays of knees, hips, elbow and neck showed dystrophic calcification. Skin biopsy demonstrated mild atrophy of the epidermis with vacuolar changes in the basal keratinocyte layer, as well as a perivascular lymphocytic infiltrate in the dermis. This leads to the diagnosis of dermatomyositis. Child was managed with steroids and physiotherapy. The child was followed up for 6 months. The condition of patient was same and lost to follow up after 6 months.

Case 2

A 12 year male child presented in the Orthopaedics (Opd) with complaints of stiffness of knees and ankle joints. The stiffness was insidious in onset and progressively increasing over 2 years. Child had difficulty in walking and squatting. Quadriceps and calf muscle wasting was present. The ankle joints were fixed in 40 degree of equinus and there was jog of movement present in both the knees. The upper limbs of the child were normal. The child had on and off episodes of low grade fever, malaise and weight loss. The patient had erythema of the mid face, skin thickening and pigmentation over

knuckles of both hands. Laboratory investigations demonstrated Hb of 8.2 g/dl, TLC of 12,600/mm³, LDH of 655 U/L, CPK of 206 U/L, CK-MB of 68 U/L and ANA was positive (Figure 2).

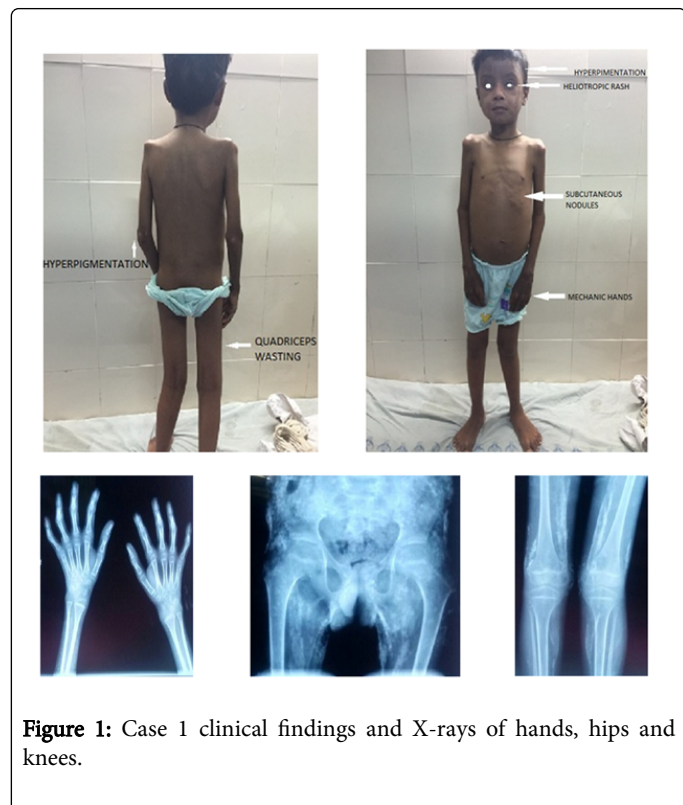


Figure 1: Case 1 clinical findings and X-rays of hands, hips and knees.

Patient was initially started on Tab Indomethacin for provisional diagnosis of myositis ossificans. But later on muscle biopsy was done which suggested interfascicular inflammatory infiltrates with muscle fiber degeneration. This leads to the diagnosis of dermatomyositis. The child was managed with prednisolone and physiotherapy. Later on surgical excision of dystrophic calcinosis around the knees was planned but parents of the child refused. The child was under the supervised follow up for 6 months. The general condition and joint stiffness improved with medication and physiotherapy.

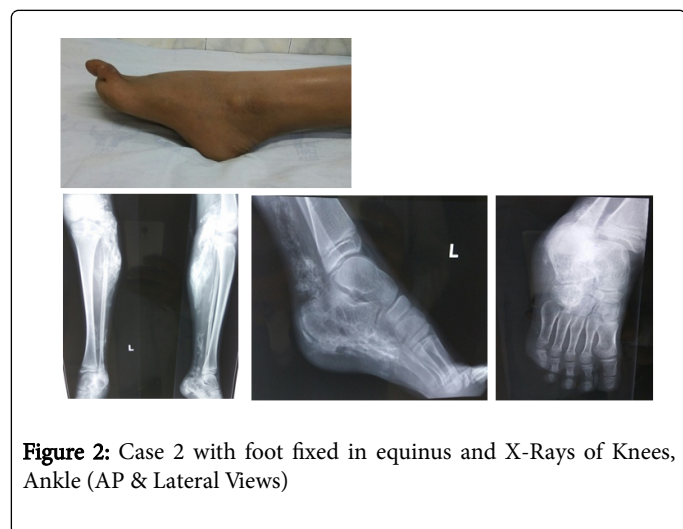


Figure 2: Case 2 with foot fixed in equinus and X-Rays of Knees, Ankle (AP & Lateral Views)

Case 3

An 11 year male patient reported to Orthopaedics (Opd) with complaints of swelling around the left hip and stiffness of both hip joints (more on left side). The stiffness progressively increased over 3 years. Child had difficulty in performing activities of daily living like squatting, climbing stairs and toilet going. Patient also complained of malaise and low grade fever. Keeping all symptoms in consideration, provisional diagnosis of Juvenile idiopathic arthritis was made. Laboratory investigations suggested of ESR of 25, C-reactive protein of 13.6 (N<6), Hb of 8.3 g/dl, TLC of 15,800. X-rays of bilateral hips depicted dystrophic calcification (Figure 3). Child further developed dysphagia and rash over the extensor aspect of joints. These symptoms and past experience leads to reconsideration of diagnosis. Further investigations demonstrated LDH of 938 U/L, CPK of 288 U/L, CK-MB of 76 U/L. Muscle biopsy suggested of perivascular and interfascicular infiltration by mononuclear cells with degenerating muscle fibers. Finally the diagnosis of dermatomyositis was considered. The child was managed with prednisolone, 0.5 mg/kg/week dose of Methotrexate and supervised physiotherapy. The child is under follow-up and currently the general condition has improved.

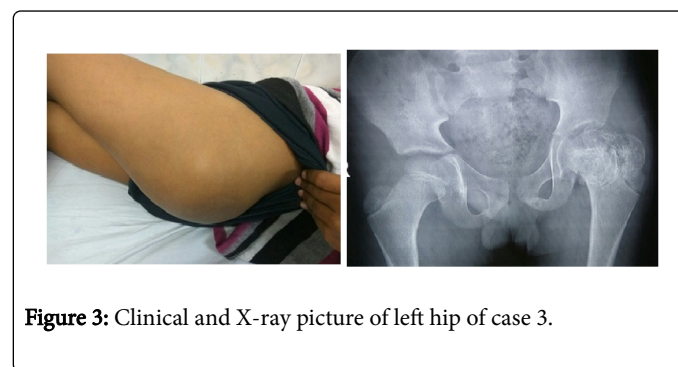


Figure 3: Clinical and X-ray picture of left hip of case 3.

Ethical committee

Informed consent has been taken from the parents of respective patients. Ethical committee clearance has been taken from the institute.

Discussion

Dermatomyositis is a rare inflammatory myopathy. It can affect individuals of all age groups, with peak incidence in adults during the fifth and sixth decades of life. The exact pathophysiology of this disease is not known but seems to be autoimmune. Some cases are felt to be paraneoplastic. Most patients present with proximal muscle weakness and rash simultaneously. Only 10% patients have muscle weakness symptoms isolatedly, whereas 30% cases have only cutaneous symptoms (dermatomyositis sine myositis). Initial cutaneous symptoms are often exaggerated by ultraviolet or sunlight exposure. The cutaneous symptoms range from pruritis, burning lesion to rash. Proximal muscle weakness involves hip and shoulder joints: patients may have complaints of difficulty standing from a sitting position or raising their arms above their heads. They also have complaints of pain and tenderness to their muscles [5]. The muscles involved in respiration and deglutition can be affected lately.

The diagnosis of dermatomyositis can be supported by serum muscle enzyme concentrations as well as autoantibody tests. Often, CK, LDH, aldolase, and aminotransferases are elevated from muscle

breakdown. Electromyography (EMG) is characterized by increased irritability with spontaneous fibrillation and sharp waves. Often, skin and muscle biopsies show inflammatory changes, segmental necrosis, or other nonspecific findings. There is no definitive test to diagnose dermatomyositis. The diagnosis is confirmed through clinical history and examination of proximal muscle weakness with skin findings and 2 of 3 laboratory criteria. These include elevated muscle enzymes, EMG changes, and tissue biopsy, as described above [5].

The symptoms are not only typical to dermatomyositis. Such cluster of clinical features can be seen in extensive calcinosis, Juvenile idiopathic arthritis, myositis ossificans progressive and chronic rheumatological disorders. Among the few cases seen in adults, calcinosis is often located in hard deposits around areas that experience frequent trauma (elbows and fingers) [6]. Socioeconomic status may play a role in the progression of calcinosis [7]. Dermatomyositis may be associated with tumors.

The cases represent the variation in signs and symptoms of dermatomyositis. The confluence of symptoms must be seen holistically otherwise misdiagnosis is common. Late diagnosis affects the long term outcome of the disease.

Treatment is based on controlling the autoimmune component of the disease. Autoimmune pathophysiology involves complement-mediated inflammation at the vascular level and direct cytotoxic effect of lymphocytes on the muscle cells.

Initial management consists of avoidance of sunlight exposure and sunscreen to prevent cutaneous lesions. High dose steroid is the mainstay of the treatment. Methotrexate, azathioprine, and mycophenolate mofetil are also common agents used in dermatomyositis. If combinations of these drugs fail, intravenous immunoglobulin can be used for short term treatment [8]. The overall prognosis for patients treated with dermatomyositis is good, with a 5-year survival rate of 95% and a 10-year survival rate of 84%. The disease may unexpectedly remit in as many as 20% of the cases and Pulmonary and cardiac manifestations are the main cause of mortality in dermatomyositis. The prognostic criteria contains persistent skin

rash and cardiac or pulmonary involvement [9]. Long term consequences are seen in 30% of patients. There may be residual weakness and contractures. Mostly patients respond to the treatment. Long-term therapy is warranted in most cases. All but one patient responded to treatment. Patients were followed till 6 months and they returned to their activities of daily living.

Conclusion

These rare case reports highlight the consideration of dermatomyositis in cases of diffuse dystrophic calcification which the orthopedicians are unaware of. This case series emphasise the importance of this condition to be known to treating clinician.

References

1. Kasteler JS, Callen JP (1994) Scalp involvement in dermatomyositis. Often overlooked or misdiagnosed. *JAMA* 272: 1939-1941.
2. Na SJ, Kim SM, Sunwoo IN, Choi YC (2009) Clinical characteristics and outcomes of juvenile and adult dermatomyositis. *J Korean Med Sci* 24: 715-721.
3. Pilkington CA, Wedderburn LR (2005) Paediatric idiopathic inflammatory muscle disease: recognition and management. *Drugs* 65: 1355-1365.
4. Pachman LM, Abbott K, Dyer A, Lipton R, Ilowite N, et al. (2006) Duration of illness is an important variable for untreated children with juvenile dermatomyositis. *J Pediatr* 148: 247-253.
5. Sontheimer RD, Costner MI, Wolff K, Goldsmith LA, Katz SI, et al. (2007) Dermatomyositis. *Fitzpatrick's Dermatology in General Medicine*, 7th edition, McGraw-Hill Professional, New York, USA.
6. Jorizzo J (2003) Dermatology. In: Bologna J, Jorizzo J, Rapini R, *Dermatomyositis*, 1st edition, Saunders, Philadelphia, 115-122.
7. Weinel S, Callen J (2004) Calcinosis cutis complicating adult-onset dermatomyositis. *Arch Dermatol* 140: 365-366.
8. Dalakas M, Hohlfeld R (2003) Polymyositis and dermatomyositis. *Lancet* 362: 971-982.
9. Benbassat J, Gefel D, Larholt K, Sukenik S, Morgenstern V, et al. (1985) Prognostic factors in polymyositis/dermatomyositis. A computer-assisted analysis of ninety-two cases. *Arthritis Rheum* 28: 249-255.