Juvenile dermatomyositis A case report

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Abstract:
Juvenile dermatomyositis (JDM) is a systemic, autoimmune inflammatory muscle disorder and vasculopathy that affects children younger than 18 years. JDM primarily affects the skin and the skeletal muscles. In the United States, the annual incidence of JDM ranges from 2.5-4.1 cases per million population. JDM primarily affects the skin and the skeletal muscles. Characteristic findings include Gottron papules, a heliotrope rash, calcinosis cutis, and symmetrical, proximal muscle weakness.

Keyword: Juvenile dermatomyositis, calcinosis cutis, inflammatory myopathy

Introduction
Juvenile dermatomyositis (JDM) is an autoimmune disorder causing vasculitis that manifests itself in children. It is the pediatric counterpart of dermatomyositis. The underlying cause of JDM is unknown. Evidence suggests a complex interplay of the innate and adaptive immune systems with environmental triggers in a genetically susceptible host. Common triggers include immunisation, infections, injuries and sunburn. JDM primarily affects the skin and the skeletal muscles. JDM is diagnosed by a combination of patient/parent observations, clinical examination and laboratory blood tests. Very often muscle biopsies and/or electromyography tests provide conclusive diagnosis. Herewith we are presenting two rare cases of juvenile dermatomyositis reported in a tertiary care hospital in Chennai.

Case report
Case-1. 7 yrs old male child, born of non-consanguinous marriage admitted with fever for 20 days duration. Edema all over the body, generalised myalgia, pain and weakness of all 4 limbs were present for 10 days duration. He is a developmentally normal child and there were no significant past or family history. On examination the child had edema with diffuse tenderness all over the body. He also had an erythematous rash over the face (picture-1) and extensor aspect of elbow and knee joints. His blood pressure was high (130/96 mm Hg). He had bilateral symmetrical proximal muscle weakness in both upper
limbs and lower limbs. Other systemic examination were within normal limits. When we proceeded with investigations, CBC, RFT were within normal limits, liver enzymes (SGOT, SGPT) were found to be elevated moderately. ESR was elevated. Serum CPK and LDH were grossly elevated (10656 IU and 1700 IU respectively). ANA and rheumatoid factor were negative. Ultrasonographic examination revealed right pleural fluid and minimal ascitis. Urine analysis showed moderate degree of proteinuria. Serum C3 was normal. Then we proceeded with EMG and muscle biopsy which showed features of inflammatory myopathy. Then we started the patient on steroids, methotrexate and folic acid. He improved symptomatically and serum CPK reduced to 300 IU. Now the child doing well on follow up.

Case-2. 6 yrs old male child Harish born to third degree consanguinous parents admitted with fever, Genaralised myalgia and weakness of all 4 limbs for 10 days duration. He also had difficulty in swallowing and nasal regurgitation for 2 days. The child was evaluated 6 months back for anaemia and jaundice, then diagnosed as autoimmune haemolytic anaemia and treated in a private hospital at Andhra Pradesh. He is a developementally normal child. There was no significant family history. On examination he was sick looking. He had genaralised edema with tenderness all over the body and erythematous rash over the cheek, knuckles, extensor aspect of elbows and knees. He also had diffuse thyroid swelling. There was bilateral symmetrical proximal muscle weakness of both upper limbs and lower limbs. On investigating the child, CBC, RFT and urine analysis were normal. ESR was elevated. LFT showed moderate elevation of liver enzymes. Serum CPK and LDH were grossly elevated (13600 IU and 2460 IU respectively). ANA and rheumatoid factor were negative, Serum C3 was normal.

X-Ray of elbow joint showed calcinosis. EMG(picture-4) and muscle biopsy were consistent with inflammatory myopathy. Thyroid auto antibodies (anti-Thyroglobulin and anti-thyroid peroxidase) were elevated. Thyroid function test showed hypothyroidism. Then the child was started on steroids, methotrexate, folic acid and L-thyroxin. The child improved symptomatically and doing well on follow up.

Discussion

Juvenile dermatomyositis (JDM) affects children younger than 18 years. JDM primarily affects the skin and the skeletal muscles. Characteristic findings include Gottron papules, a heliotrope rash, calcinosis cutis, and symmetrical, proximal muscle weakness. Although similar in many respects to adult dermatomyositis with characteristic skin findings and muscle weakness, JDM is often associated with calcinosis cutis, cutaneous ulcerations, and vasculopathy characterized by intimal proliferation of small blood vessels and infarctions(1,2). Traditionally, diagnosis of JDM has been based on the following 5 criteria, specified by Bohan and Peter in 1975 (3,4). These are characteristic skin rash, proximal muscle weakness, elevated muscle enzymes, myopathic changes on electromyography, abnormal muscle biopsy findings Typical skin findings in combination with 3 other criteria are necessary to make the diagnosis. Patients with the characteristic rash who fulfill only 2 criteria have probable JDM. On the basis of a 2006 international consensus survey, expanded criteria have been proposed(5). These in part reflect the development of noninvasive techniques since the 1975 criteria were formulated and include the typical findings on muscle MRI and ultrasonography, nailfold capillaroscopy.
abnormalities, calcinosis, dysphonia. The current model of the pathogenesis of JDM involves both humoral and cell-mediated mechanisms that cause vascular and muscle damage. Autoantibodies directed against an unknown endothelial antigen may cause vascular injury, resulting in ischemia and subsequent muscle damage with increased expression of major histocompatibility complex (MHC) class I and II(6,7,8,9,10,11).

Immune complex deposition mediates vascular injury, resulting in activation of complement and muscle inflammation(9,11,12,13). Emerging research suggests that type I interferon-alpha/beta inducible genes of the innate immune system play a central role in the pathogenesis of dermatomyositis(11). The etiology of JDM is incompletely understood. Seasonal clustering of JDM in the months of April and May suggests the role of environmental triggers in the onset or exacerbation of the disease(14). Infectious agents include viruses, parasites, and bacterial antigens that may produce a break in self-tolerance. Infectious agents implicated include coxsackie B virus, parvovirus B19, enteroviruses, streptococcus species(15,16). Constitutional, respiratory, and GI symptoms may occur within 3 months of onset of juvenile dermatomyositis (JDM). Muscle involvement can be insidious, symmetrical proximal muscle weakness involving the deltoids, quadriceps, or both is a prominent clinical finding in JDM(15). Other common symptoms include fever, dysphagia, dysphonia or hoarseness, myalgias, arthralgias, abdominal pain, and melena from GI involvement as a consequence of vasculopathy(15). A malar rash in a photosensitive distribution with sparing of the nasolabial folds (heliotrope rash) may occur, making the diagnosis of JDM difficult to distinguish from systemic lupus erythematosus. Periorbital edema is sometimes present Erythematous, violaceous scaly plaques (Gottron papules) may occur on the extensor surfaces of the extremities. Sparing of the interphalangeal spaces is observed. Nailfold telangiectasias, periungual erythema may be seen. Hypertrophic, ragged cuticles may accompany periungual erythema(17). Inadequately treated children have persistent nailfold abnormalities reflective of skin disease activity but not muscle involvement(18). Diffuse vasculopathy may be associated with vasomotor instability, such as Raynaud phenomenon, livedo reticularis, or vascular infarctions on the medial canthus of the eyelids. Mechanic's hands may occur, with hyperkeratosis and peeling of the skin over the lateral and palmar aspects of the fingers(19).

Laboratory studies
Laboratory studies in the workup of juvenile dermatomyositis (JDM) include an erythrocyte sedimentation rate (ESR), muscle enzyme levels, lupus profile, and myositis-specific antibody assays. The ESR is commonly elevated in patients with JDM, but this finding is nonspecific. Levels of muscle enzymes such as aspartate aminotransferase, lactate dehydrogenase, creatine kinase, and aldolase may be elevated early in the disease course. Nailfold capillary microscopyapillary loss and formation of bushy loops representing capillary dilatation and branching. An elevated ANA level may be seen in approximately half of patients with JDM(17). Generally, the extractable nuclear antigens (SSA, SSB, Sm, RNP, DNA) are negative. Muscle ultrasonography reveals increased muscle echogenicity, attenuation, and reduced bone surface echo. These changes are not specific for JDM, however, and this technique is not widely used(20).
Magnetic resonance imaging (MRI) with T2-weighted fat suppression and short tau inversion recovery (STIR) is useful in the diagnostic workup because it reveals edema, which is a marker of muscle inflammation. Electromyography (EMG) reveals a reduction of the motor unit action potentials in the proximal muscles and fibrillation potentials suggestive of fiber splitting, necrosis, and vacuolization. However, the EMG findings may be normal in approximately 19% of children. A muscle biopsy is not usually performed to confirm the diagnosis of JDM, as it is for adult myositis. However, it is needed in the workup of juvenile polymyositis. If muscle biopsy is performed, a moderately weak muscle should be selected with specimens frozen for cryostat sections to perform histologic and enzyme histochemical stains and immunocytochemistry for major histocompatibility complex (MHC) antigens, immunophenotyping of T cells, and detection of cytokines and complement.

Treatment

A multidisciplinary approach is required to prevent and reduce long-term morbidity in juvenile dermatomyositis (JDM). For active muscle disease, oral corticosteroids are the mainstay of treatment. High doses (1-2 mg/kg/day) are initially used until improvement or for 4-6 weeks, with a slow taper to avoid relapse. Weaning of steroids often occurs over 1-2 years. For refractory or severe disease, pulse therapy with intravenous methylprednisolone is used at a dose of 30 mg/kg, with a maximum dose of 1 g daily for 3 days. Calcium and vitamin D supplementation is recommended for patients on long-term corticosteroid therapy. Second-line agents are routinely added for steroid-sparing effects and for recalcitrant or refractory disease. Methotrexate has been the most widely accepted agent and is used at doses of 10-20 mg/m² per week orally or subcutaneously with 1 mg/d of folic acid supplementation. Methotrexate administration may be started at the outset of severe disease (eg, moderate to severe weakness, manifestations of vasculopathy) or started if patients fail to respond to high-dose corticosteroids within 6 weeks. The use of methotrexate in conjunction with a tapering course of prednisone may be effective in reducing the long-term cumulative dose of corticosteroids. Cyclosporin A has been used as an alternative, effective, steroid-sparing agent in JDM. Preliminary data also suggest that the use of cyclosporine A in combination with methotrexate may be associated with further improvement in clinical outcome. Intravenous immunoglobulin (IVIG) may also be used in patients with JDM who are steroid-resistant or steroid-dependent at a dose of 1 g/kg on 2 consecutive days, or 2 g/kg in one day, then every month thereafter, generally for a 6-month course. Other second-line agents include azathioprine and mycophenolate mofetil. Biologic agents such as the anti–tumor necrosis factor agents have had mixed results. More recently, infliximab has shown major clinical benefit in 5 patients with refractory JDM (21). Another promising agent for JDM, although still in clinical trials, is the anti-CD20 monoclonal antibody rituximab (22). Various therapies are used to treat skin manifestations of JDM. Photosensitive rashes in patients with JDM may be exacerbated by sunlight exposure; sun avoidance, and judicious use of sunscreens that protect against ultraviolet A and B rays. Topical corticosteroids are used to treat skin manifestations of JDM. Hydroxychloroquine at a dose no greater than 5
-7 mg/kg/day may be beneficial. Topical tacrolimus (0.1%) has been used to treat cutaneous disease, but this use is falling out of favor. Methotrexate, which is an established second-line agent for muscle involvement in JDM, may also be effective for cutaneous manifestations. Patients should be followed on a regular basis, with monitoring of muscle enzymes and muscle strength every 3-6 months. Remission occurs when stable improvement or normalization of muscle strength and muscle enzymes is observed over 6 months. Rehabilitation plays an active role in the first-line management of myositis to restore and maintain muscle strength and endurance and to prevent contractures. If the myositis is active, then passive range of motion exercises are recommended; pool therapy may also be helpful. When the disease is stabilized, isometric and isotonic exercises have been recommended. However, one study indicated that an intensive muscular training program involving resistance training may be beneficial and safe without worsening of muscle inflammation (23). For patients with severe contractures, physical and occupational therapy with assistive devices is recommended.

References


picture 1-Heliotropic rash over the face
Picture 2-Gottrons patches over the extensor aspects of interphalangeal joints Picture 3 -Gottrons patches over elbow joint

Picture 4 -EMG report showing evidence of inflammatory myopathy