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CASE REPORT

Juvenile Dermatomyositis

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ABSTRACT

Juvenile dermatomyositis (JDM) is an important subtype of dermatomyositis characterized by inflammation of muscle, skin and gastrointestinal tract. A 14-year-old girl, with a history of fever, joint pain, easy fatigability and a rash since the age of 3 years is described. Physical examination, laboratory evaluation, electromyography (EMG) and muscle biopsy were suggestive of a chronic inflammatory process involving the muscles, most likely dermatomyositis. The report highlights the importance of a muscle biopsy as the gold standard for diagnosing dermatomyositis.

Key words: *Juvenile dermatomyositis. Muscle biopsy. Arthritis. Rash. Adolescent female. Gower's sign.*

INTRODUCTION

Dermatomyositis is an idiopathic inflammatory myopathy with characteristic skin manifestations. Although the disorder is rare, with a prevalence of one to 10 cases per million in adults and one to 3.2 cases per million in children, early recognition and treatment are important ways to decrease the morbidity of systemic complications.[1]

In 1975, Bohan and Peter first suggested a set of criteria to aid in the diagnosis and classification of dermatomyositis and polymyositis (PM). Four of the 5 criteria are related to the muscle disease, as follows: progressive proximal symmetrical weakness, elevated levels of muscle enzymes, an abnormal finding on electromyography, and an abnormal finding on muscle biopsy. The fifth criterion was compatible with cutaneous disease.[2]

Recently, a scoring tool was proposed for muscle biopsy evaluation in patients with juvenile dermatomyositis, which was based on four domains of change: inflammatory, vascular, muscle fiber, and connective tissue. The inflammatory and muscle fiber domains had the highest reliability and agreement.[3]

We report here a case of an adolescent female child with muscle biopsy proven dermatomyositis.

CASE REPORT

The patient, a 14-year-old adolescent, was in her usual state of health till 3 years of age when it was noticed that she had difficulty in walking and running. Her milestones otherwise had been achieved at appropriate ages, while the child continued to have spikes of low grade intermittent fevers. She was taken to various doctors and had received treatment for about a year now, which included various topical applications for the rash and multi vitamin supplements for her stated 'weakness'.

Physical examination revealed an emaciated girl with fever of 38.4°C. There was a discoid, erythematous, violaceous rash involving the dorsum of her hands and fingers. Bilateral depigmentation and dermal thickening of her knuckles were present. A generalized erythema involving the shoulders and the back was also present.

Joint and muscular system examinations revealed a bilateral asymmetric involvement of the proximal muscles of the lower limbs. She demonstrated difficulty in standing from a sitting position and vice versa. Gower's sign was hence found to be positive. Examination of the upper limbs revealed permanent contractures (Figure 1). Rest of the systemic examination was unremarkable.

Laboratory examination showed elevated creatine kinase (337 mg/dl), lactate dehydrogenase (2229 mg/dl) and aldolase (8.7 mg/dl), with normal levels of serum aminoaspartate, serum alanine aspartate, C-reactive

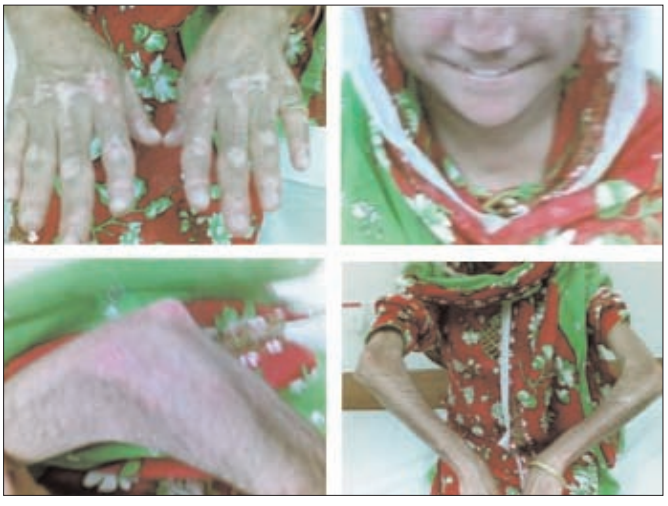

Figure 1: A 14-year-old girl with characteristic rash and joint deformity.

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protein and erythrocyte segmentation rate. Autoimmune work up revealed a positive rheumatoid factor (RF), and anti-nuclear antibody (ANA) with a homogenous pattern (titres not available) with a negative anti-smith antibody, anti-mitochondrial antibody and complement (C3, C4 and C1q) levels. Urinalysis results were within normal limits.

Electromyography (EMG) was abnormal, with indications of muscle irritability. It was reported to have short-duration, low-amplitude, polyphasic units and increased spontaneous activity with fibrillations, complex repetitive discharges, and positive sharp waves with early recruitment, highly consistent of a myopathic process.

Muscle biopsy was taken from vastus lateralis muscle. The histopathology showed marked variation in the size, with focal and perifascicular atrophy and loss of muscle fibers. Scattered fibers showed splitting and internalization. Dense, non-specific inflammatory infiltrate was observed, pre-dominantly centred around the vessels and hence was stated to be consistent with a chronic inflammatory process, most likely dermato-myositis. The raised alkaline phosphatase levels were said to signify a primary muscle disease. Figures 2 (a-d) detail the patient's skin and muscle biopsies.

There was marked clinical improvement after she was started on prednisone and hydroxychloroquine therapy. Her laboratory parameters from last follow-up have been reported to be within the normal range. She is in remission now. She is still on the same therapy, but on maintenance doses.

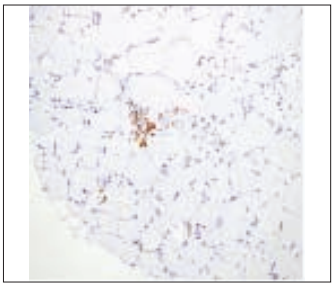

Figure 2a: LCA immunostain highlights the lymphocytes around muscle fibers.

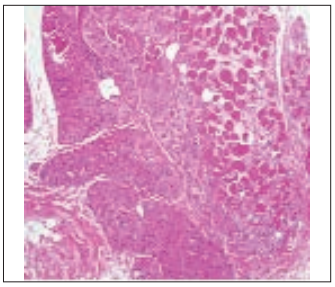

Figure 2b: Skeletal muscle tissue (4X) with variation in fiber size and hypereosinophilic fibers.

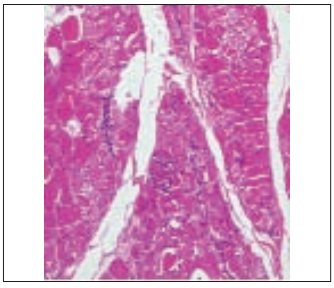

Figure 2c: Skeletal muscle (20X) fascicles showing perifascicular atrophy and lymphoid infiltrate.

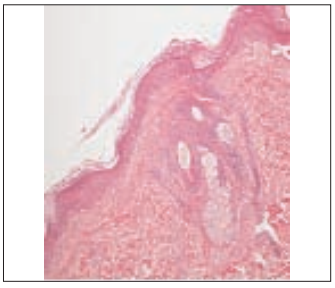

Figure 2d: Skin biopsy (10X) showing mild lymphocytic infiltrate in the dermis.

DISCUSSION

Although dermatomyositis was first described in 1863, its course is so variable as to be difficult to predict. In children the clinical pattern tends to be more characteristic than in adults, in whom vascular lesions have been less characteristic of the pathological findings. The average age at diagnosis is 40 years and almost twice as many women are affected as men.

Juvenile dermatomyositis (JDM), an important subtype of dermatomyositis is a multisystem disease characterized by inflammation of muscle, skin and gastro-intestinal tract. In children, immune complex vasculitis of various severity is the pathological hallmark. By definition, the onset of JDM occurs in children younger than 16 years of age, with girls being affected more than boys.[4]

Dermatomyositis presents with characteristic cutaneous and joint manifestations. These manifestations of dermatomyositis are generally grouped as patho-gnomonic, characteristic, compatible, less common and rare.

Among the cutaneous findings, 'heliotrope', a macular rash with periorbital edema, is considered a characteristic finding of dermatomyositis, as are periungual telangiectasias.[5] The rash occurs early in the course of the disease in 30 to 60 percent of patients.[3] The characteristic lesions of the shawl sign and the V-sign appear as erythematous, poikilodermatous macules distributed in a "shawl" pattern over the shoulders, arms and upper back and in a V-shaped distribution over the anterior neck and chest. Mechanic's hand may be associated with an increased risk of interstitial lung disease.[6]

The rash over the patient's hands was consistent with Gottron's papules, the pathognomonic joint manifestation of JDM, which by definition are violaceous papules overlying the dorsal interphalangeal or meta-carpophalangeal areas, elbow or knee joints, occur in approximately 70% of patients with dermatomyositis.[7]

Patients may exhibit weakness of the truncal muscles that requires them to use their arms to push themselves up from a prone position i.e. Gower's sign. The sign was present in this patient.

JDM usually can be diagnosed on a clinical basis in children with a characteristic rash and proximal muscle weakness. However, a complete history should be obtained and a physical examination performed, including a thorough review of systems, with an emphasis on myositis-related presentations and evidence of skin changes.

The routinely done laboratory work up includes creatine kinase (CK), lactate dehydrogenase (LDH), aldolase, and aspartate aminotransferase (AST) for the evaluation

of myopathy. Elevation in the serum concentration of one or more of these enzymes occurs sometime in nearly every patient.[8]

Muscle biopsy is the definitive test in establishing the diagnosis of JPM or JDM and in excluding many other causes of myopathy. The hallmark of histologic change in JDM is the presence of a vasculopathy in skin, muscle, and gastrointestinal tissue that progresses in stages.[9] In this patient, the diagnosis was based on the muscle biopsy which suggested inflammatory changes consistent with dermatomyositis.

The goal of the therapy is to improve function and prevent disability. The treatment regimen must be instituted early and requires a team approach between the physical therapist, dermatologist and family physician. Of the many therapeutic measures employed at one time or another for juvenile dermatomyositis, corticosteroids have received the most attention. In those individuals with a good response to initial glucocorticoid treatment, muscle enzymes should return to normal in 6 to 12 weeks, followed by the return of muscle strength. The use of IVIG in JDM has been evaluated in several preliminary studies involving children who had previously failed treatment with glucocorticoids, and in most cases immunosuppressive agents.[10]

Poor prognostic indicators in juvenile dermatomyositis are late onset of treatment, initial treatment with a dosage of prednisone that is too low, recalcitrant disease and pharyngeal involvement.

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