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CASE REPORT

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Juvenile Dermatomyositis in a Nigerian Girl: a Case Report.

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Summary A case of Juvenile dermatomyositis (JDM) in a 10 year old Nigerian girl is herein reported to discuss some of the features of the disease and challenges in management of such a rare but crippling autoimmune vasculopathy of childhood. She was referred to the University of Maiduguri Teaching Hospital (UMTH) with an eight-month history of recurrent fever, abdominal pain, and a four-month history of body rash and inability to walk or sit. Muscle biopsy and clinical findings consistent with JDM were found. Her condition improved with steroids, cytotoxic therapy and physiotherapy. Some investigation and treatment modalities could not be accessed for the benefit of the patient. Although, the outcome of patients with JDM has improved with the discovery of steroids, the disease is shown to have a variable course, with attendant social and financial implications especially to the immediate family.

Introduction

Juvenile dermatomyositis (JDM) is the most common childhood inflammatory myopathy.[1] It is a systemic, autoimmune inflammatory muscle disorder and vasculopathy that affects children younger than 18 years, primarily affecting the skin and the skeletal muscles. Characteristic findings include Gottron papules, a heliotrope rash, calcinosis cutis, and symmetrical proximal muscle weakness.

An estimated annual incidence of 3.4, 3.3 and 2.7 cases per million children were reported in whites, blacks and Hispanics respectively,[2] and a female to male ratio of 2.3:1 and 5:1 in the United States and United Kingdom respectively.[2,3] The median age of onset of JDM is 6.8 years in girls and 7.3 years in boys, with a median diagnosis delay of 3-4 months.[4] Prior to the advent of corticosteroids, the prognosis was poor. Brunsting and Banker types of JDM have been described in children, with the former type characterized by slow progression and better outcome compared to the latter type characterized by rapid onset of symptoms and gastrointestinal tract involvement.[5,6]

We report a case of JDM in a Nigerian girl which to the best of our knowledge has not been previously reported in Nigeria probably because of its rarity or because it was usually missed by clinicians. Some of the features disease and challenges encountered in management are also highlighted.

Case report

HUA was a 10 year old primary school girl referred from a general hospital to University of Maiduguri Teaching Hospital (UMTH) with an eight month history of recurrent fever, abdominal pain, and facial puffiness, four months history of body rash, dysphonia and inability to walk or sit. Prior to referral, she was being treated at the referring facility for acute glomerulonephritis (AGN) and anaemia, where she had received blood transfusion and other medications without improvement. Although, a history of sore throat preceding the onset of

symptoms was obtained, no history of change in urinary frequency, volume or colour was found.

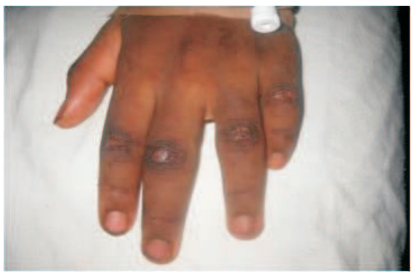
There was also no history of haematuria or dark coloured urine. The body rash was more on the upper torso, extensor surfaces of both upper and lower limbs and the buttocks. No associated joint swelling or pain but there was generalised body ache with classical history of proximal muscle weakness that Progressed to inability to walk, sit-up and hold the neck. At about the same time, she was noticed have nasal quality speech with occasional regurgitation of fluid via the nose. She had no preceding history of exposure to vaccines, drugs or insect bite. HUA was not a known sickle cell disease patient and had no history suggestive of diabetes mellitus. There was no family history of such or similar presentation.

On admission, physical examination revealed a chronically ill looking pale, girl with facial puffiness but was afebrile, not jaundiced and had no significant peripheral lymphadenopathy. Her weight was 26kg (80% of expected for age) and her length was 137cm (100% of expected for age). The main findings were in the musculoskeletal system which revealed generalised body tenderness. She was unable to raise her arms even to feed herself. She was curled up almost in a foetal position, unable to sit and had some contractures at the elbow and the knee joints.

Maculopapular skin eruptions were observed in the upper torso, extensor surfaces of arms and legs and the buttocks. A crop of peri-orbital eruptions was noted below the right medial canthus (heliotrope) and the skin over the metacarpal and proximal interphalengeal joints fitting the pattern of typical Gottron papules.

The lesions were hypertrophied and erythematous

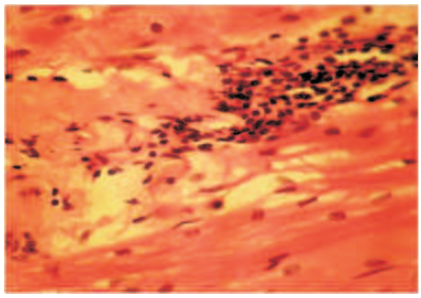
Fig 1



Although, no subcutaneous mass or swelling was found, an ulcer on the right elbow measuring 2x3cm in diameter with lobulated floor was noted. Diagnosis of Juvenile Dermatomyositis (JDM) was entertained. Blood pressure and urinalysis remained normal throughout the period of admission. HIV antibody test and Rheumatoid factor were negative. Antistreptolysin O titre was positive (400 IU) but total creatine-kinase (CK) titer was within normal limits.

Skeletal muscle CK iso-enzyme analysis was not done due to non availability of the reagents. Other immunologic tests such as anti-nuclear antibody, myositis associated autoantibody were not done due unavailability in our centre. Stool for occult blood, urine microscopy, culture and sensitivity were negative. Complete blood count and serum electrolytes, urea and creatinine were not remarkable. Histology of incisional muscle biopsy from the right upper thigh demonstrated focal areas of inflammation, necrosis, few myofibre fragmentation as well as atrophy and fibrin which are in keeping with JDM (Fig 2).

Fig 2



Photomicrograph showing myofibre necrosis, atrophy and lymphocytic infiltrates: MG X 200). Radiograph of the upper arms showed no evidence of calcifications.

She was commenced on prednisolone 2mg/kg/24hr and four weeks later, weekly methotrexate 15mg/m^2 was added because of slow response to the steroid. Following resolution of muscle pain and tenderness, physiotherapy was commenced. The patient responded to the treatment with resolution of the fever; headache, muscle pain and desquamation and healing of skin eruptions. She regained ability to feed herself and was able to sit unsupported for up to an hour. About a week after initial improvement, she developed high grade continuous fever and anorexia. Sepsis work up was done, but no organism was isolated and no focus for the infection was found. She was given anti-malarial and antibiotic medications to no avail. HUA subsequently died two weeks following the onset of the fever.

Discussion

The diagnosis of JDM is challenging especially in a developing country like Nigeria where adequate facilities, equipment and expertise may be lacking. In 1975, Bohan and Peter[7] proposed criteria for the diagnosis of JDM. These criteria include characteristic skin rash, proximal muscle weakness, elevated muscle enzymes, myopathic changes on electromyography and abnormal muscle biopsy findings. Typical skin findings in combination with

Three other criteria are necessary to make a definitive diagnosis. Patients with the characteristic skin eruptions who fulfill only two criteria are adjudged to have probable JDM. The proposed criteria have been expanded to include typical MRI and ultrasonography findings, including affected muscle, nail fold capillaroscopy, presence of calcinosis, and dysphonia. The index patient had fulfilled almost all the above criteria except the tests that could not be done due to lack of necessary facilities. The symptoms, signs and skin eruptions found in the patient were typical of JDM. The median time for diagnosis after onset of the disease was put at four months,[1,4] diagnosis was made 8 months into the illness in our patient. The delay in diagnosis was due to late presentation, referral, and lack of suspicion of the disease, due to its rare occurrence.

Although the cause of JDM is not known, infection-triggered autoimmunity has been proposed as a possible precipitating factor. Infectious agents implicated include coxsackie B virus, parvovirus B19, enteroviruses, and *Streptococcus* species.[1,4] Preceding history of sore throat was obtained in the patient, so also elevated ASO titre. It is probable therefore that a streptococcal infection triggered the autoimmunity in the patient. Noninfectious agents implicated in the onset of JDM include D-penicillamine, vaccinations, and bone marrow transplants among other triggers.[4]

Negative rheumatoid factor and anaemia are common occurrence in patients with JDM.[1,4] Although, an elevated skeletal muscle creatinine kinase (CK) is an important finding in patients,[1,7,8] a normal level does not exclude JDM as the enzyme level is usually elevated in the early phase of the disease, during the period of maximum muscle inflammation and destruction.[1] Pachman et al[9] reported that two out of five patients with definite JDM had normal muscle enzyme when re-evaluated some months later, though they had elevated enzyme levels initially at the time of diagnosis. The patient presented to us eight months into the illness and had a normal total CK. It is possible that she had elevated CK in the early phase of the disease consistent withthe natural history of JDM. Other muscle enzymes That may be elevated in JDM are aspartate aminotransferase, lactic dehydrogenase and aldolase.

Radiographic MRI using T2 weighted images and fat suppression localizes the active site of disease for diagnostic muscle biopsy and electromyogram, which are each non diagnostic in 20% of instances if Not directed by MRI.[1] The typical muscle biopsy pathological finding[10] of JDM in the patient despite

The fact that the biopsy was not guided by MRI probably suggest extensive muscle involvement in the patient. Other evidence of severe disease in the patient include nasal speech and regurgitation of liquids through the nose and gastrointestinal tract symptoms.[1,8]

Calcifications occur in 20-40% of children with dermatomyositis and is implicated in increasing the morbidity and mortality of the disease.[11] In JDM, calcifications are more frequent at anatomic sites that are normally exposed to daily minor trauma but not usually mineralized, such as the elbows or behind the knees.[9] Calcinosis on the other hand, is seen as crusted papules or plaques around joints or as non-healing sores. The intractable ulcers at the elbows in the patient may suggest the existence of calcinosis. Sometimes, the calcified material is extruded through the skin as a white cheesy exudate, leaving behind a dry pitted scar.[8] Muscle calcification results in contractures or severe muscular pain. Calcinosis is thought to be dystrophic, as damaged muscles release mitochondrial calcium into matrix vesicles that promote mineralization. Calcinosis lesions are rarely present at diagnosis but are usually found later during the course of the disease.[1,5] Delayed treatment and severe disease have been reported as risk factors for development of calcinosis.[1,4,8]

The goals of treatment include control of inflammation manifesting as pain, muscle weakness, skin lesion and prevention and management of short term and long term complications of the disease and treatment. The main stay of therapy is steroids. For severe and refractory cases, intravenous methyl prednisolone can be given. Second line drugs methotrexate, cyclosporine, azathioprine and hydroxychloroquine. Cyclosporin and hydroxychloroquine are reported to be effective especially for skin involvement.[1,8] Intravenous immunoglobulin (IVIG) may be indicated by relapse, incomplete response, or for steroid sparing purposes,[8] the addition of the other second line drugs in the management of JDM also has the benefit of steroid sparing effect.

Due to the effect and contribution of cytokine like TNF-α in the pathophysiology of dermatomyositis, the use of biologic agents in management of JDM is currently gaining grounds. The use of anti-TNF-α has shown mixed results in the management of JDM.[4,8] Rituximab, a monoclonal CD20+B cell-depleting antibody, is another potential promising new biologic agent in the management of JDM, its potential use is also investigated in a broad range of conditions involving B-cells.[4,8]

Treatment adjuncts include calcium and vitamin D to correct the osteopenia of JDM and to decrease the frequency of bone fracture, avoidance of exposure to sun and also use of sunscreen, to provide protection against ultraviolet A and B.[1,8] Antacids or H2 blockers, if dyspeptic symptoms are precipitated bysteroids and antibiotics, especially for an infected ulcer. Vitamin supplements may also be beneficial. Physiotherapy provides passive stretching early inthe disease and once active inflammation has resolved, direct reconditioning of muscles to regain strength and range of motion is initiated. Bed rest is not indicated. Social work services may help facilitate adjustment to the frustration of physical impairment in a previously active child. Formation of social support groups as in other chronic debilitating illnesses may assist children with this condition and their parents or care givers.The course of JDM may be as brief as eight months

With complete recovery, or it can last two or more years with a continuing requirement for treatment.[8] Acute exacerbations and remission without any stabilization of the initial course of the disease, late progression with a recurrence of active disease after a prolonged remission have been reported.[8]

The period of active symptoms has decreased from about 3.5 years to less than 1.5 years with more aggressive immunosuppressive therapy and a mortality of about 1-10% was reported in western literature.[1,4,5] Prior to the advent of corticosteroids, it was reported that one third of affected children died and another one third were disabled.[1] The outcome of treatment of JDM in Nigeria is not known. Although HUA had initial clinical improvement, sepsis set in (but cultures were sterile) and scuttled the management of the patient towards achieving complete remission.

Even though, the health burden of rare disease conditions like JDM on the Nigeria nation may be low, its effect and burden on the affected family is high, with attendant social and financial implications. The challenges posed by this disease to care givers especially in developing countries like Nigeria are enormous. These concerns are depicted by this case report.

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