Possible juvenile dermatomyositis in a 7-year-old Nigerian girl: a case report

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Abstract

Juvenile Dermatomyositis (JDM) is a systemic autoimmune disease that presents in children before their 16th birthday. Although the skin is the most obvious organ affected, it also affects the skeletal muscles, the lungs, the gastrointestinal system and the heart. The paucity of rheumatologists, especially paediatric rheumatologists, and dermatologists in Nigeria results in delayed identification and management. This is a case report of a 7-year-old Nigerian girl who presented with clinical and laboratory features of possible juvenile dermatomyositis. Limitations encountered during management of this patient were the absence of electromyogram and muscle biopsy for histology. It is the hope of the authors that this case will heighten the index of suspicion and add to the growing literature of JDM in Nigeria and Africa at large.

Key words: Idiopathic inflammatory myopathy, Juvenile dermatomyositis, Proximal muscle weakness, Helioptrope rash and Gottron's lesion

Introduction

Idiopathic Inflammatory Myopathies (IIM) are a group of disorders which are characterized by symmetric proximal muscle weakness, and elevated muscle specific enzymes. A subtype of IIM presents with additional characteristic skin lesions. Juvenile Dermatomyositis (JDM) is the commoner of the two forms of Idiopathic Inflammatory Myopathy (IIM) occurring in children. It is a rare chronic autoimmune disease with onset in childhood, before the 16th birthday[1]. Although elevated muscle enzymes are a major feature of IIM, this may not be seen in JDM.

Case report

A 7-year-old primary school pupil, who presented to the rheumatology clinic of University of Benin Teaching Hospital, with a complaint of widespread rashes of one year duration. The informant was her aunt who had brought her from the village on account of this complaint. Rashes had begun as erythematous and edematous lesions located on the peri-orbital regions of both eyes. They were described also as non-tender and non-pruritic. She subsequently developed other rashes on her neck, anterior chest wall, both elbows, knuckles of the hands and over her knees. She had associated hair loss, and poor growth with delayed eruption of primary dentition. There was also a history of pain and swelling in the right wrist with resultant flexion deformity as well as complaints of tiredness and difficulty rising up from a squatting position. On account of these, she had been withdrawn from school by her parents.

Examination revealed a young child who was ill looking, with obvious heliotrope rash in both peri-orbital regions (Figure 1) and alopecia in both parietal regions of the head (Figure 2). She also had presence of V-sign, Sleeve sign, Gottron's lesions on knuckles of the hands (Figure 3) and scaly rashes over the knees. (Figure 4). She had Grade 3 power in the proximal muscles of both upper and lower limbs with positive Gower's sign. There was swelling of the right wrist joint, flexion deformity and tenderness of the joint.

Figure 1: Heliotrope rash

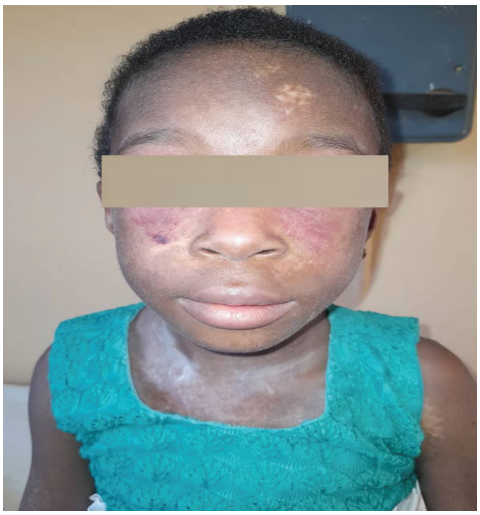


Figure 2: Alopecia

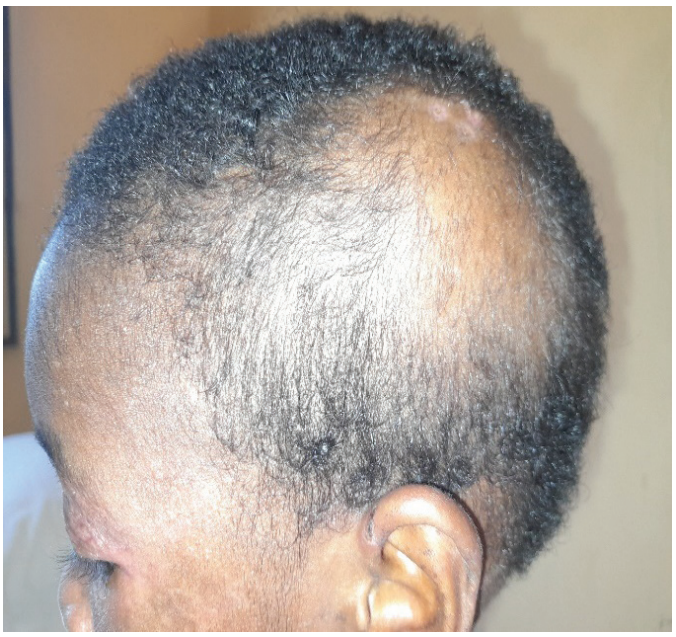


Figure 3: Gottron's lesions

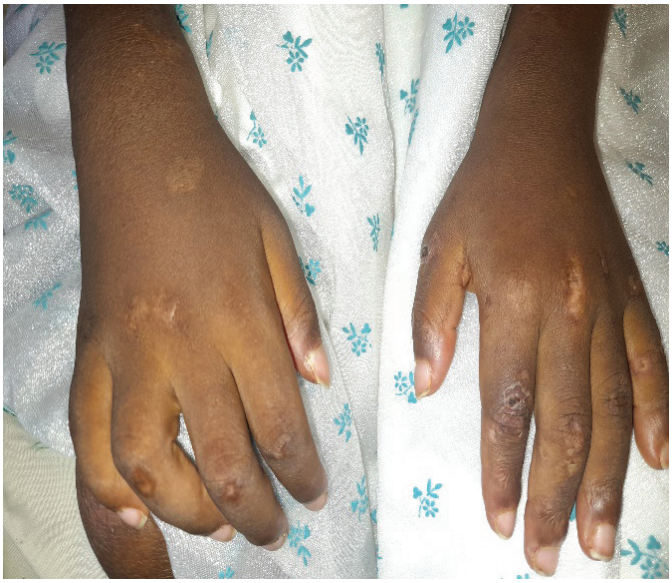
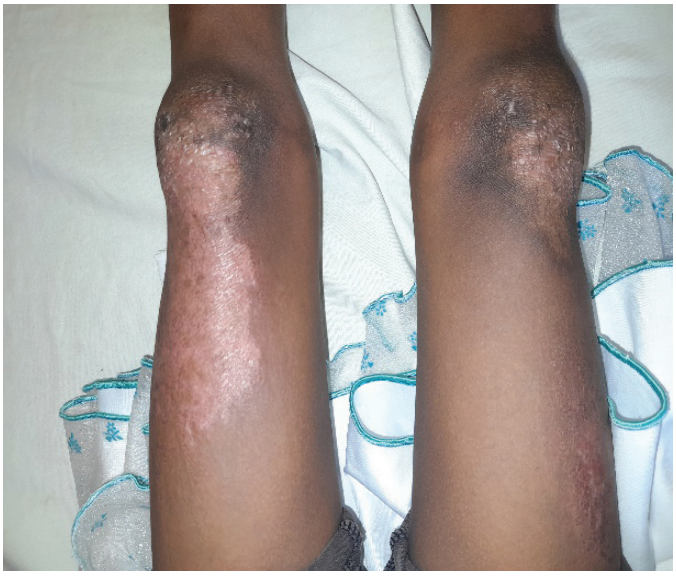


Figure 4: Scaly plaques over both knees



Investigation results revealed normal muscle enzymes: creatine kinase 107 U/L (15 – 170), aldolase 4.86 U/L (0 – 7.6), aspartate aminotransferase 34 IU/L (5 – 34) and alanine aminotransferase 16 IU/L (0 - 55). She had a white blood cell count of 6.0 x 10^9/L, red cell count of 3.92 x 10^{12}/L, haematocrit of 33.8% and platelet count of 201 x 10^9/L. Hepatitis B and C, and retroviral screening were negative. Anti-nuclear antibody was positive with a 1:640 titre (speckled pattern). Anti-ds-DNA and anti-Smith antibodies were negative while serum complement levels were within normal range. Screening for myositis autoantibodies was also negative. This test screened for the following; antibodies against Jo1/histidyl tRNA synthetase, Ku, Mi-2, PL7, PL12, EJ, OJ, signal recognition particle (SRP), Ro-52 and PM/Scl.

A diagnosis of possible juvenile dermatomyositis was made using the Bohan and Peter diagnostic criteria. Criteria met included; presence of characteristic rash, and symmetrical proximal muscle weakness. She was commenced on subcutaneous methotrexate at 10mg once a week, with a weekly titration upwards to a target dose of 1mg/kg weekly. She was also commenced on oral prednisolone at 0.5mg/kg/day, and calcium/vitamin D3 supplementation. Sun protective measures and physiotherapy were also instituted. The patient is currently doing well, with no report of new skin lesions, improvement in muscle power and has since resumed school.

Discussion

Juvenile Dermatomyositis (JDM) is the commoner of the two forms of Idiopathic Inflammatory Myopathy (IIM) occurring in children, with juvenile polymyositis being the other rarely encountered entity. It is a rare chronic autoimmune disease with onset in childhood, before the 16th birthday[1]. It is characterized by well recognized rashes and symmetrical proximal myopathy. It needs to be differentiated from infectious and neuromuscular disorders which present with similar complaints of progressive weakness in childhood. The diagnosis of JDM is usually made using the Bohan and Peter diagnostic criteria, which was developed for adult DM but has been adapted for the paediatric age group. There are attempts at developing diagnostic criteria for JDM and JPM[2,3].

The estimated annual incidence of JDM is 2.5 – 4.1 per million children per year in the United States[4]. There have been few reported cases in sub-Saharan Africa. Adelowo *et al*[5], reported a case of an 11-year-old Nigerian girl with JDM while Grijsen *et al*[6] reported that of a 4-year-old Kenyan girl. Okongo'o *et al*[7] had done a nine-year cross-sectional study in South Africa where a 1.8:1 female to male ratio was found amongst twenty-five cases. In this study, most patients were of indigenous African descent (52%) followed by mixed race and those of European ancestry. A cohort study done in Egypt found the mean age at onset of disease to be 5.9±2.8 years[8].

A diagnosis of possible JDM was made in the index patient based on the presence of cutaneous manifestations of heliotrope rash, V-sign, Sleeve sign, Gottron's lesion and alopecia in addition to the presence of symmetrical weakness of the limb girdles. Due to unavailability of investigative tools, an electromyogram was not done while her guardian declined muscle biopsy which is the gold standard for detecting muscle inflammation. T2 weighted Short Tau Inversion Recovery (STIR), Magnetic Resonance Imaging (MRI) also detects the presence of inflammation as hyperintensities but is not specific.

JDM differs from JPM not only by the presence of cutaneous lesions but also by histologic findings. In JDM, CD4+ T helper cells predominate in the perimysial and perivascular regions and MHC class I expression is limited to damaged muscle fibers in the perifascicular regions while in JPM, CD8+ T cytotoxic cells predominate in the endomysial region and MHC class I is up regulated virtually in all fibers regardless of the presence of inflammation.

The heliotrope rash was the most common cutaneous manifestation of JDM seen in the study by El-Garf *et al*[8] Juvenile dermatomyositis differs from adult dermatomyositis by the presence of systemic features like fever, and a more rapid progression of the cutaneous lesions including the development of calcinosis. This may be attributed to the presence of anti-TIF1-gamma antibody[9]. Calcinosis was found to be present in 40% of patients in the study done by Okongo'o *et al*[7] and children of African ancestry had a higher prevalence (62%) of calcinosis compared to patients of other ancestry. Muscle enzymes like creatine kinase may not be elevated in JDM as seen in adult DM. The occurrence of cancer has also not been frequently associated with JDM as seen in adult cases[9]. The course of JDM may be monophasic, polyphasic or chronic, with the presence of Gottron's papules and nail fold abnormalities early in the disease, and is associated with a longer time to remission[10].

Our patient was managed using the Children Arthritis and Rheumatology Research Alliance consensus treatment plan C with daily oral prednisolone and weekly subcutaneous methotrexate[11] with resultant improvement in her clinical condition. She was also commenced on sun protective measures and physiotherapy. Ciclosporin, azathioprine, hydroxychloroquine and mycophenolate mofetil are other Disease Modifying Rheumatic Drugs (DMARDs) that can be used in management of JDM. Cyclophosphamide may be used in cases of skin ulcerations and gastrointestinal perforation. Biologic DMARDs such as infliximab and rituximab as well as abatacept, Janus kinase inhibitors and intravenous immunoglobulins also have roles in the management of JDM[12].

This case report aims to increase the awareness of JDM and other IIM in African children amongst general practitioners, paediatricians and physicians alike. Early recognition and referral improve the outcome of patients.

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