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Case Report

Unravelling the Diagnostic Dilemma of Juvenile Scleroderma: A Case Report

Obichukwu NG[1], Ofiaeli OC[1,2], Mbachu CNP[1,2]

[1]Department of Paediatrics, Nnamdi Azikiwe University Teaching Hospital Nnewi, Anambra state, Nigeria

[2]Department of Paediatrics, Faculty of Medicine, College of Health Sciences, Nnamdi Azikiwe University, Anambra State, Nigeria.

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Abstract:

Juvenile scleroderma (JS) is a rare and often overlooked connective tissue disease in Paediatric populations. Its diagnosis is particularly challenging due to its multi-systemic involvement, and in developing countries like ours, the lack of diagnostic tools further complicates timely identification. We present the case of an 11-year-old male who presented to our children's emergency room with a 2-year history of progressive skin thickening, flexion contracture and immobility of the right thumb. There was generalized body pain and swelling of the right upper limb. Additionally, the patient experienced recurrent cough, night sweats, easy fatigability, and occasional palpitations, though without significant weight loss.

After an initial misdiagnosis and several ineffective treatments, the suspected diagnosis of JS led to a trial treatment with methylprednisolone and methotrexate, to which the patient responded favourably. This case underscores the importance of a high index of suspicion for the prompt diagnosis and management of rare childhood rheumatic diseases, as delay in diagnosis can exacerbate morbidity and increase mortality.

Introduction

Juvenile scleroderma comprises of many connective tissue disease with a known hallmark of skin fibrosis.[1] Scleroderma is derived from Greek words skleros (hard or indurated) and derma (skin). [2] Hippocrates firstly described it as thickened skin condition.[3] In 1945, scleroderma was noted to be a systemic disease and progressive in nature, hence was previously referred to as progressive systemic sclerosis.[4] Scleroderma is very rare in children and presents a problem in diagnosis when it occurs with difficult long term management.[5] Its third most frequent Paediatrics rheumatic disease after Juvenile idiopathic arthritis and systemic lupus erythematosus.[6] There are two major types thus: Juvenile localized scleroderma, seen in majority of cases, is mainly limited to the skin and the systemic sclerosis, seen in about 10% of cases, which is known to have multi-systemic organ involvement like the lungs, heart, kidney, digestive system, skin etc. [1,7-9] The aetiology of JS is unknown, though both environmental and genetic factors involving the immune system dysfunction of the vascular tissue and extracellular matrix have been demonstrated.[1,10] The incidence of JSS ranges from 0.27 to 2.9 cases per million children per year.[10] The mean age of onset is 8 to 11 years, the females are more affected than males. No known racial predominance.[10]

The clinical manifestation of JSc is usually insidious with prolonged periods between onset and diagnosis. Majority of children present with skin changes like tightening, thinning and atrophy of the hand with raynoud's phenomenon. [4,7,11] Other associated presenting complaints are arthritis, althralgia, muscle weakness, muscle pain, subcutaneous calcification, dysphagia, dyspnea and palpitation. The definitive diagnosis of JSc is made when a child has the major criteria; that is proximal sclerosis/induration of the skin and any 2 of the minor criterias.[1] The minor criterias includes the presence of cutanous sclerodactyly, raynauds phenomeno/telangiectasias, dysphagia or gastric reflux, arrhythmia or heart failure, renal pathology, respiratory system affectation, neurologic like neuropathy, musculo-skeletal pathology and positivity of any of the antinuclear antibody serologic screening.[1,10]Although JSc is rare, it is one of the most severe childhood rheumatologic conditions. We reported here a rare case of Juvenile Scleroderma in an eleven-year-old patient.

Case Report

An eleven-year-old male patient presented with 2-year history of stiff right hand, muscle pain of the whole right hand, difficulty in lifting the right hand, recurrent cough with easy fatigueability. Child also had heart burn, difficulty in swallowing and occasional palpitation. For these symptoms child was treated for tuberculosis and suspected malignancy in the first visited hospital for over 6weeks. With worsening symptoms, mother decided to bring child for expert care. The initial clinical findings on presentation revealed weight 51kg which was above the 50th percentile for age, height was above the 50th percentile for age and gender. The Blood pressure was at 90th percentile for age, height and gender. Pulse rate was 82

beats per minute, full volume and regular. Respiratory rate was 22 cycles per minutes and SPO2 was 98% in room air. Marked tenderness was noted on the whole right upper limb with classical skin thickening with stiffness and flexion contracture of the metacarpo-phalangeal joint of the right thumb. Moderate epigastric tenderness was noted. Other systemic examination findings were normal. The blood biochemistry, complete blood count and urinalysis were normal. The rheumatoid factors were negative, viral serologic test for hepatitis B, HCV and HAV were negative. MRI of the right hand showed an inflammation of the extensor tendon, dermal biopsy demonstrated increased connective tissues under the sweat glands which was consistent with scleroderma. The electrocardiography was unremarkable, though the Echocardiography was consistent with constrictive pattern of carditis. No ocular uveitis was detected. Esophageal reflux scintigraphy was positive. Respiratory function test was normal. Serum electrolytes urea and creatinine and complete blood counts were normal. No pathology was detected on abdominal ultrasound. Tuberculosis screening tests like, Monteux test, AFBX 3, gene xpert, chest x-ray were all negative. No blast cells were detected from the peripheral blood film test. On immunological assessment, the antinuclear antibody test and anti-Scl-70 antibody were positive, though anti double stranded DNA test was negative. The acute phase reactants like the ESR, CRP were elevated.

The child was diagnosed with Juvenile systemic scleroderma due to sclerotic involvement of the finger and the typical findings on the dermal biopsy and MRI of the the right arm and fingers. Intravenous methyl prednisolone 30mg/kg every other day for one week was given with weekly intravenous methothrexate for 8 consecutive weeks, then followed up with oral methotrexate and prednisolone. Child was also enrolled to physical exercise programme of the right upper limbs. Child was in remission by 2nd month of treatment. Currently child is symptom free but still being followed up.

Discussion

JSc is a rare severe autoimmune disease known to be severe and life threatening autoimmune disease with multiorgan inflammation causing fibrosis. [11] The fibrosis following deposition of collagen and other macromolecules of connective tissue on the skin and multiple internal organs following numerous humoral and cellular immunologic abnormalities and severe fibro proliferative alterations in the microvasculature. [12] The etiology remain unknown though some speculations with increasing evidence suggests interaction between the environment and some genetic factors that triggers micro vascular injury leading to structural and functional endothelial cell abnormalities. This is true for the index case as the child presented with stiff right arm with marked tenderness, classical skin thickening and flexion contracture of the metacarpo-phalangeal joint of his right thumb. This findings were in consonance with similar cases reported by Danaoui et al,[11] , Ahmad et al,[13], Faleye et al,[6] and Irene et al.[14] This shows that JSc is usually associated with musculoskeletal affectations, hence inclusion of skin manifestations as major criteria, which must be present for the definitive diagnosis of JSc.[10,12]

Apart from the musculoskeletal system affectation, the index case also had affectation of the internal organs/systems as evidenced by the occasional palpitation, recurrent cough and difficulty in swallowing, most probably from gastroesophageal reflux with fibrosis of the gastrointestinal tract. This findings was also noted in similar case reported by Santos et al,[7] Goel et al,[5] and Danaoui et al.[11] This portrays the multi-systemic nature of JSc though some reported cases of JSc had only skin manifestations.[13] This buttress the fact that JSc may not necessarily affect the internal organs for a definitive diagnosis to be made, hence lowering the risk of delayed diagnosis. It is important to note that while ANA test and acute phase reactant tests like ESR, CRP were elevated in the index case and other reported cases of JSc with systemic manifestations,[9,15] Faleye et al and Ahmad et al noted that they were normal in a case of linear JSc.[6,13] This shows that further studies may be needed to establish or substantiate this findings.

The index case responded well to use of steroids and methotrexate, which was noted in similar case reports. [5,15] Furthermore, same result was obtained with the use of similar therapy for JSc with skin manifestations.[13] Though Ferguson et al reported successful treatment of recalcitrant linear JSc with use of infliximad and leflunomide.[16] This shows that use of infliximad and leflunomide should be considered in any JSc patient that cannot tolerate the side effects from use of steroid.

The index case was reported to enlighten the physicians of the possibility of mis – diagnosing a case of JSc in sub-region. This report will help to heighten the clinicians' index of suspicion for JSc in any child that presents with obvious skin manifestations. Since research have shown that early diagnosis and intervention is associated with good response and prognosis, while delayed diagnosis and treatment of JSc can lead to permanent squeal with significant impact to quality of life.[9]

Conclusion

We have presented a case of JSc that had skin manifestations with involvement of internal organs/systems. This case was successfully treated with 8 weeks course of steroid using methylprednisolone and methothrexate. Currently child has been on follow-up with resolution of the clinical signs and symptoms. High index of suspicion is needed for early diagnosis and intervention with rare childhood rheumatic disease like JSc.

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