A Child in Horse-Riding Stance

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A 6-year-old girl presented with progressive tightness of skin over pelvic and shoulder girdles resulting in stiffness of extremities giving her a fixed horse-riding stance [Figure 1]. It started as an ill-defined stony hard area noticed on the lateral side of right buttock at 2 years of age and there was a gradual progression of skin induration over proximal extremities. There was exaggerated lumbar lordosis, outer aspects of both the buttocks were flat and hollowed, and lower back and lumbar region showed mild hypertrichosis [Figures 1 and 2]. Skin on the involved body parts was rock-hard and not pinchable. Face, neck, hands, and feet were not involved. There was no history of Raynaud's phenomenon, arthralgia, dyspnea, or dysphagia. Hemogram, ESR, random blood sugar, renal function, and urinalysis were within normal limits. Antinuclear antibody test was negative. A deep biopsy was taken from an involved area of her left thigh including skin and underlying fascia.

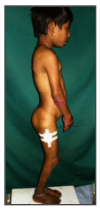

Figure 1
Horse-riding stance of the child

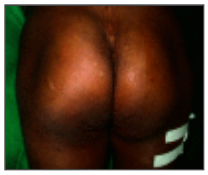

Figure 2
Hollowed contour of the buttocks with mild hypertrichosis of the lumbo-sacral region

Histopathological findings

There were (H and E) unremarkable epidermis, sparse, perivascular lymphocytes in the upper dermis and thick, sclerotic collagen bundles in the reticular dermis. There was no evidence of appendageal distortion. Subcutaneous fat and fascia were normal in appearance in the biopsied specimen.

Question

What is your Diagnosis?

Answer

Diagnosis: Stiff skin syndrome

Stiff skin syndrome (SSS) is a rare disorder encountered in childhood causing fixed contracture deformity of the joints. There is no underlying musculo-skeletal or visceral involvement.[1] Esterly and McKusik described this disorder in 1971[2] and thereafter Jablonska described a distinct subset of the disease, “congenital fascial dystrophy.”[2] Description of SSS is heterogenous and present consensus is that it is a triad of rock-hard induration of skin, hypertrichosis and restricted joint mobility.[3]

SSS manifests in early childhood or any time by 7 years of age. It involves body areas with abundant fascia, around pelvic and shoulder girdles and thoracic wall.

SSS is insidious in onset and either advances slowly or remains non-progressive. Joint contractures develop secondary to involvement of overlying fascia and skin. Some patients may develop a tiptoe gait and barrel chest deformity due to extensive fibrosis.[1] Diagnosis is based mainly on classical clinical presentation as there is no pathognomonic histopathological feature. In the variant designated as congenital fascial dystrophy, approximately four to six-folds thickening of the involved fascia is seen. There is a dense collection of coarse and giant collagen fibrils (amianthoid/asbestoid fibers). Fascial involvement was not demonstrable in the biopsied specimen in our patient; however, there was deposition of thick collagen in the reticular dermis. This finding is similar to that of Liu *et al*.,[1] in all their six reported cases.

As evidenced by histopathological and immunopathological studies, SSS is a non-inflammatory fibrotic disorder, without any vascular hyper-reactivity.[1] Pathogenesis of SSS is not well understood. Loeys *et al*.[4] have reported that SSS is caused by mutations in the sole Arg-Gly-Asp sequence-encoding domain of fibrillin-1. Microfibrill, an ordered polymer of fibrillin-1, helps in elastic fiber assembly and activation of the profibrotic cytokine transforming growth factor-β (TGF-β). Altered cell–matrix interactions in SSS result in excessive microfibrillar deposition, impaired elastogenesis, increased TGF-β concentration, and signaling in the dermis.

Various other disorders encountered in childhood may simulate SSS.[1] These include systemic sclerosis, generalized morphea, eosinophilic fascitis, scleredema of Buschke, and nephrogenic fibrosing dermopathy. Rare inherited disorders such as mucopolysaccharidosis, lysosomal storage disease, juvenile hyaline fibromatosis and Hutchinson-Gilford progeria may present with focal or diffuse thickening of skin, subcutaneous nodules and flexion contracture of joints.[1] Typical clinical features of the disease, initiating during early childhood as woody induration of skin around pelvic and shoulder girdles, sparing face and distal extremities and progressive stiffness of large joints without any systemic involvement are diagnostic of SSS.

Restricted mobility is the factor impairing quality of life in SSS. Long term physiotherapy helps in rehabilitation and some of the affected patients have been reported to develop skills in sports and given child-birth.[5] Our patient was advised active and passive physiotherapy for limb joints to improve mobility. However, she was lost for further follow up. Extensive involvement of the thoracic fascia may lead to restricted costal mobility leading to respiratory insufficiency and subsequent death.[5,6] Pages *et al*.[6] have reported a patient with SSS, who suffered from acute abdomen due to extensive necrosis of the intestinal wall and died subsequently. Such symptoms or any respiratory symptom should be given urgent consideration in these patients as overlying rigidity of the parietal wall may make it difficult to assess the extent of underlying illness.

Footnotes

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Conflict of Interest: Nil.

Article information

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