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Case Report

Congenital presentation of linear morphea in a 4 year old boy: A rare case report with literature review

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Abstract Linear morphea is a variant of morphea characterized by linear induration of the skin, often associated with pigmentary changes. It is common in children in their first or second decade of life. Congenital morphea is extremely rare, and it usually presents as a linear variant affecting the extremities or face. There have been only 25 cases reported so far. In this report, we present another rare case of congenital linear morphea in a 4-year-old boy. The child was started on topical tacrolimus and targeted ultraviolet B phototherapy.

Key words
Congenital, linear morphea.

Introduction

Morphea, a chronic autoimmune inflammatory disease, is characterized by sclerosis of the skin.[1] Current guidelines classify morphea into 5 subtypes that include circumscribed, generalized, linear, pansclerotic and mixed subtypes.[2] Linear morphea is a morphological variant of morphea commonly seen in children, often affecting the extremities and face.[1] 90.0% of affected children are between 2 and 14 years of age. The etiology is unclear, but an interplay between genetic, environmental, and autoimmune factors may result in its pathogenesis.[3] Linear morphea affects not only the skin but can also extend to the underlying subcutaneous tissue, muscle, and bone, resulting in functional impairment and cosmetic problems. Many children develop extracutaneous complications such as severe atrophy of the extremities, deformities, contractures, and limb length discrepancies.[4,5] Congenital morphea is extremely uncommon, with only 25 cases published in the literature so far.[6]

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Case report

A 4-year-old boy was brought to our Dermatology outpatient department by his parents with complaints of itchy hypopigmented to brownish linear lesions over the right thigh and groin area since birth. The lesion was initially smaller in size, which then gradually progressed and attained the present size. There was no history of any birth trauma. He had no other associated systemic complaints and his personal or family history did not reveal any Raynaud's phenomenon or connective tissue disease. On examination, multiple well-defined hypopigmented/ brownish atrophic plaques with scaling and crusting surrounded by lilac-coloured borders were seen arranged in a linear pattern extending from the right lower thigh to

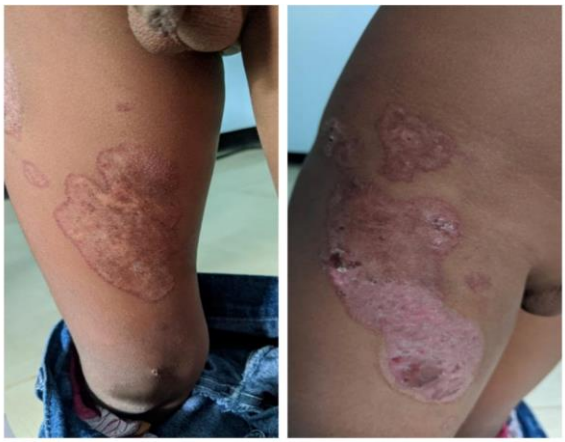

Figure 1 Multiple well-defined hypopigmented to brownish atrophic plaques with scaling and crusting and typical 'lilac borders' arranged in a linear pattern extending from the right lower thigh to groin.

the groin, with the largest plaque measuring 12×5 cm and the smallest one of size 2×1 cm **(Figure 1)**. The overlying skin was indurated, difficult to pinch with hairs sparsely scattered over the plaque. No arthritis, joint contracture, or limb shortening was observed. Clinical possibility of morphea was considered and a deep punch biopsy including subcutaneous fat was done, which revealed typical features such as full thickness hyalinisation of the dermis, perivascular lymphocytic collections, and atrophy of sebaceous glands with sparse sweat glands, confirming our diagnosis **(Figure 2&3)**. Anti-nuclear antibody and rheumatoid factor tests were negative, and complete blood analysis, urine analysis, renal and liver function parameters were all within normal limits. The child was started on targeted ultraviolet B phototherapy and topical tacrolimus 0.03% ointment.

Discussion

Scleroderma is an autoimmune connective tissue disorder comprising both morphea and systemic sclerosis. Both types represent either ends of the spectrum.[7] The pathogenesis of morphea is unknown, but genetic, autoimmune, and environmental factors that cause microvascular

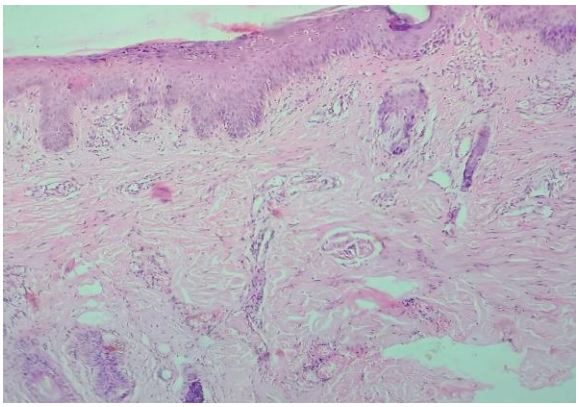

Figure 2 Dermis showing full thickness hyalinisation with abnormal collagen bundles and absence of sebaceous glands and sparse swear glands (H&E×10x).

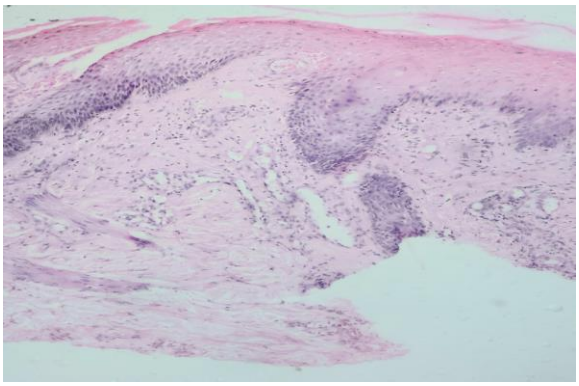

Figure 3 Dermis showing perivascular lymphocytic infiltrates and replacement by hyalinised collagen bundles (H&E×40x).

injury, leading to an imbalance between collagen synthesis and degradation, are thought to be important mechanisms.[3] Zulian and Laxer[2] classified morphea in children into five types, which included circumscribed, linear, generalized, pansclerotic and mixed types.

Linear morphea is characterized by linear indurated plaques associated with pigmentary changes, often involving the limbs, face, and scalp, or sometimes the trunk. The plaques sometimes follow the course of Blaschko's lines. A linear lesion involving the frontal or fronto-parietal area with or without hemifacial atrophy is known as en coup de sabre variant of morphea. Parry Romberg syndrome comprises hemifacial atrophy without dermal sclerosis. Lesion crossing over the joint can result in

significant disability and deformity and also may involve the underlying subcutaneous fat, muscle, or bone, causing severe atrophy of the affected limb.[8] Linear morphea affecting the limbs can lead to musculoskeletal complications such as arthralgia, arthritis, joint contracture, severe limb atrophy, limb shortening, and gait problems. In the en coup de sabre type, neurological involvement such as seizures, migraine, trigeminal neuralgia, behavioural problems, and learning dysfunction can occur. Eye abnormalities seen are loss of eyebrows and eyelashes on the affected side, anterior uveitis, enophthalmos, and myopathy of the eye muscles.[8,9] Associated oral and dental abnormalities include tooth malocclusion, bone asymmetry, temporomandibular joint involvement, overgrowth of the anterior lower third of the face, and hemiatrophy involving the ipsilateral tongue.[10] Congenital morphea is extremely rare and a highly underestimated condition in neonates, with only a few cases reported in the literature. Diagnosis is often missed for several years, owing to the ignorance about this condition. Its early appearance may be mistaken for other common conditions such as port-wine stain and nevus simplex.[6] The first case of biopsy proven congenital morphea was reported by Joshi *et al.*[11] in a 4-year-old boy over the abdomen. Castanon *et al.*[12] observed a similar case in a 6-month-old girl. A study by Zulian *et al.*[13] among 750 cases of juvenile localized scleroderma reported 6 (0.8%) cases with scleroderma lesions at birth. All these patients had linear scleroderma, in which en coup de sabre type was seen in 4 cases. They also observed a delay in diagnosis of up to 4 years. Recently, Tomaszewska[5] and colleagues described a case of linear morphea profunda in a boy that was misdiagnosed several times before being finally confirmed as morphea at the age of 7. Lipson and colleagues[14] described a congenital case in which the diagnosis was not reached until 36 years of age. Monsour *et al.*[6] performed a multicenter retrospective cohort study between 2001 and 2016, combining it with a comprehensive search of past literature, and identified 25 unique cases of congenital morphea. The most common subtype observed was linear morphea in 19 patients, of which 13 had linear morphea of the head and 5 had linear limb involvement. Four out of five patients (80.0%) with linear morphea of the extremities had limb deformity or musculoskeletal impairment. The higher percentage of linear morphea among those with congenital morphea supports the theory that linear morphea occurs as a result of genetic mosaicism or a developmental anomaly, as the skin lesions appear to follow the Blaschko's lines.[15] In view of the high complications associated with linear morphea, early diagnosis and prompt initiation of treatment with subsequent periodic monitoring is imperative to limit the severity of extracutaneous complications.

Positive antinuclear antibodies are seen in 47-67% of cases with linear morphea, and anti-Scl70 antibodies, a marker of systemic sclerosis, may be found in 2-3% of cases with morphea. Eosinophilia and polyclonal hyper-gammaglobulinemia are found in the active stages of the disease.[14,16] However, none of these tests are sensitive or specific for morphea, nor do they correlate with disease activity. Rheumatoid factor is seen in 16% of patients and is significantly associated with arthritis.

Deep skin biopsy, including the subcutaneous fat, aids in the diagnosis. The early inflammatory phase of morphea is characterized by dermal infiltrates of lymphocytes, eosinophils, plasma cells, and mast cells with excessive collagen deposition. In the advanced sclerotic phase, dermis appears thickened and hypocellular with increased collagen, sparse inflammatory infiltrate, and atrophic eccrine glands, which appear higher as a result of

excessive collagen replacing the subcutaneous fat. Blood vessels are decreased in number with thick vessel walls and narrowed lumens.[9] In our case, the child is probably in the sclerotic phase as suggested by the histopathological findings.

The current recommendation for the treatment of moderate to severe morphea is a combination of methotrexate and systemic steroids as an initial "bridge therapy". Other treatment options in resistant disease are mycophenolate mofetil, ciclosporine, and hydroxychloroquine. Phototherapy along with topical agents is useful in superficial morphea, but interference with the school schedule is a major factor limiting its regular use in children. Topical treatment options such as steroids, steroid-calcipotriol combination, or tacrolimus are useful in localized disease.[9,17]

Conclusion

We have reported this case of linear morphea due to its extreme rarity and the unique presentation of it since birth. Congenital morphea with linear morphea in particular can cause severe, disabling extracutaneous complications if left untreated early. Hence, a high index of suspicion for this condition in neonates and infants and continuous observation are of vital importance to minimize the complications in such cases.

References

1. Cyrus N, Jacobe HT. Morphea and lichen sclerosus. In: Wolff K, Goldsmith LA, Katz SI, Gilchrest BA, Paller AS, Leffell DJ. Fitzpatrick's Dermatology in General Medicine, 8th ed.,New York: McGraw Hill, 2012. p. 1106-1117.
2. Laxer RM, Zulian F. Localized scleroderma. Curr Opin Rheumatol 2006;18:606-613.
3. Fett N, Werth VP. Update on morphea: part I. Epidemiology, clinical presentation, and pathogenesis. *J Am Acad Dermatol*. 2011;**64**:217-28.
4. Zulian F. Systemic manifestations in localized scleroderma. *Curr Rheumatol Rep*. 2004;**6**:417-24.
5. Tomaszewska KA, Krzysiek J, Kobusiewicz A, *et al*. Congenital morphea profunda in a 7-year-old male patient with coexistence of fibrous hamartoma of infancy: a rare case report. *Postepy Dermatol Alergol*. 2018;**35**:638-40.
6. Mansour M, Liy Wong C, Zulian F, *et al*. Natural history and extracutaneous involvement of congenital morphea. Multicenter retrospective cohort study and literature review. *Pediatr Dermatol*. 2018;**35**:761-8.
7. Dimitrova V, Yordanova I, Gospodinov DK. Generalized morphea: a case report. *J IMAB*. 2007;**13**:68-70.
8. Li SC. Scleroderma in children and adolescents: Localized scleroderma and systemic sclerosis. *Pediatr Clin North Am*. 2018;**65**:757‑81.
9. George R, George A, Kumar TS. Update on management of morphea (Localized Scleroderma) in children. *Indian Dermatol Online J*. 2020;**11**:135-45.
10. Trainito S, Lorenzo F, Martini G, et al. Odontostomatologic involvement in juvenile localised scleroderma of the face. *J Paediatr Child Health*. 2012;**48**:572‑6.
11. Joshi A, Al-Mutairi N, Nour-Eldin O. Congenital skin lesions presenting as morphea in a 4-year-old. *Pediatr Dermatol*. 2006;**23**:94-5.
12. Castañón CS, Martínez Blanco J, Rodríguez Díaz E. Congenital morphea. *An Pediatr (Barc)*. 2010;**73**:64-65.
13. Zulian F, Vallongo C, de Oliveira SK, *et al*. Congenital localized scleroderma. *J Pediatr*. 2006;**149**:248-51.
14. Lipson J, O'Toole A, Kapur S. Delay in diagnosis of congenital linear scleroderma. *J Cutan Med Surg*. 2015;**19**:156-8.
15. Weibel L, Haper JI. Linear morphoea follows Blaschko's lines. *Br J Dermatol*. 2008;**159**:175-81.
16. Torok KS. Pediatric scleroderma: systemic or localized forms. *Pediatr Clin North Am*. 2012;**59**:381-405.
17. Zulian F, Culpo R, Sperotto F, *et al*. Consensus‑based recommendations for the management of juvenile localised scleroderma. *Ann Rheum Dis*. 2019;78:1019‑24.