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The Vanishing Face: Clinical Presentation of Hemifacial Atrophy and Its Dental Implications

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Abstract: ***Hemifacial atrophy is a rare, acquired, neurocutaneous syndrome characterised by a progressive but self-limiting atrophy of the skin and subcutaneous tissue on one side of the face. Early diagnosis and a multidisciplinary approach are essential to prevent worsening of functional and aesthetic complications. We report the case of a 13-year-old male presenting with unilateral facial atrophy accompanied by firm, hyperpigmented plaques aligned along Blaschko's lines on the right side of the face. Diagnostic evaluations, including serology and imaging, confirmed the diagnosis of linear morphea en coup de sabre. Histopathological analysis revealed dermal fibrosis with lymphocytic infiltration. Dental examination identified irreversible pulpitis in the lower right first permanent molar, attributed to skeletal and dental asymmetry. Root canal treatment was performed, followed by prosthetic rehabilitation using a custom 3D-printed zirconia endocrown. Concurrently, the patient received topical tacrolimus as part of dermatologic management. This case highlights the significance of early recognition and coordinated care in patients with linear scleroderma involving craniofacial structures. Dental practitioners play a vital role in managing oral complications and preserving function and aesthetics in such patients. Multidisciplinary collaboration is essential to improve outcomes and quality of life.***

Keywords: Blaschko lines, Progressive facial hemiatrophy, Morphea, CAD/CAM Endocrown

1.Introduction

Scleroderma is a connective tissue disorder that primarily affects the skin and mucous membranes, characterized by excessive collagen deposition in the dermis and subcutaneous tissues. This leads to thickening of these layers, eventually resulting in scar-like lesions. The condition presents in various forms, including morphea (a localized variant), as well as subtypes such as plaque morphea, bullous morphea, linear scleroderma (including morphea en coup de sabre [ECDS] and progressive hemifacial atrophy [PHA]), and systemic sclerosis (also known as systemic scleroderma).[1,2]

Morphea, also known as juvenile localised scleroderma,[3] can lead to lasting cosmetic and functional consequences, such as areas of hyperpigmentation or hypopigmentation, superficial or deep tissue atrophy, and reduced joint mobility or deformities.[2,3] Unlike systemic sclerosis (SSc), morphea does not exhibit features such as sclerodactyly, Raynaud's phenomenon, or internal organ involvement. All forms of morphea share a common characteristic—excess collagen production by fibroblasts in the affected tissues—though the exact mechanism behind fibroblast activation remains unclear. Potential factors contributing to the development of morphea include endothelial cell damage, immune and inflammatory responses, and abnormal regulation of collagen synthesis. The presence of autoantibodies in many patients also supports an autoimmune origin.[4] It is at least ten times more common in children compared to pediatric systemic sclerosis. The estimated incidence is between 0.4 and 1 per 100,000 individuals, with a female-to-male ratio ranging from 2 to 3:1.[3,4]

Several classifications have been proposed to classify morphea, including that by Laxer and Zulian [5] (Table 1), which consists of 5 subtypes:

Table 1: Preliminary Proposed Classification of Juvenile Localized Scleroderma

Main Group	Subtype	Description
Circumscribed morphea	Superficial	Oval or round circumscribed areas of induration limited to epidermis and dermis, often with altered pigmentation and violaceous, erythematous halo (lilac ring). They can be single or multiple.
	Deep	Oval or round circumscribed deep induration of the skin involving subcutaneous tissue extending to fascia and possibly muscle. Lesions may be single or multiple. Sometimes subcutaneous involvement occurs without skin changes.

Linear scleroderma	Trunk/limbs	Linear induration involving dermis, subcutaneous tissue, and sometimes muscle and bone. Affects the limbs and trunk.
	Head	Includes En coup de sabre (ECDS), affecting face and scalp, sometimes involving muscle and bone; and Parry Romberg syndrome (progressive hemifacial atrophy), involving unilateral loss of tissue on one side of the face.
Generalized morphea		Induration starting as four or more plaques (≥3 cm) that become confluent and involve at least two of seven anatomic sites (head–neck, limbs, trunk).
Panclerotic morphea		Circumferential involvement of limb(s), affecting skin, subcutaneous tissue, muscle, and bone, without internal organ involvement.
Mixed morphea		Combination of two or more of the previous subtypes, described by their predominant representation (e.g., mixed morphoea [linear-circumscribed]).

Another frequently cited classification is that by Peterson et al,[6] which also includes 5 subtypes:

Plaque morphea

- Morphea en plaque
- Guttate morphea
- Atrophoderma of Pasini and Pierini
- Keloid morphea (nodular morphea)
- Lichen sclerosus et atrophicus

Generalized morphea
Bullous morphea
Linear morphea

- Linear morphea (linear scleroderma)
- En coup de sabre
- Progressive hemifacial atrophy

Deep morphea

- Subcutaneous morphea
- Eosinophilic fasciitis
- Morphea profunda
- Disabling pansclerotic morphea of children

This article aims to report and analyze a rare pediatric case of linear morphea en coup de sabre with dental manifestations, emphasizing the importance of interdisciplinary management in preserving oral function and facial aesthetics.

2.Case Report

A 13-year old male patient presented to the outpatient department of Pediatric and Preventive Dentistry, University College of Medical Sciences and Guru Teg Bahadur Hospital, with a chief complaint of pain in his lower right back tooth region for 12 days. On cutaneous examination, the patient exhibited features consistent with linear scleroderma (morphea), including unilateral facial atrophy and hyperpigmented indurated plaques along the lines of Blaschko, primarily involving the right side of the face [Figure 1]. These are embryonic cell migration pathways often unmasked in cutaneous mosaicism and inflammatory dermatoses. In this patient, the lesions predominantly involved the right side of the face in a distribution suggestive of linear morphea en coup de sabre.

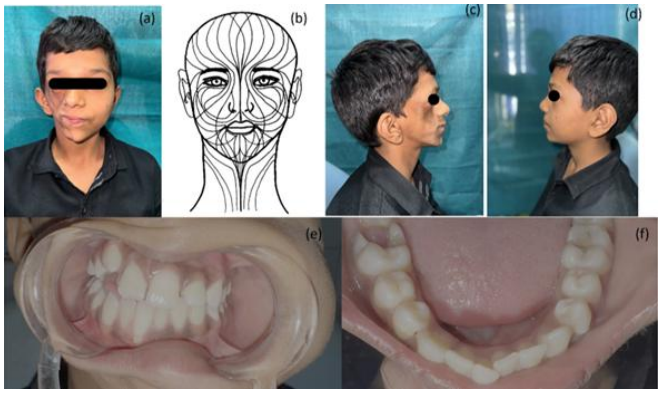

Figure 1: (a) Frontal facial view showing facial asymmetry, atrophy, and linear depression (en coup de sabre appearance) on the right side of the face, typical of linear morphea; (b) Blaschko lines; (c) Right lateral profile showing significant soft tissue depression and midface underdevelopment; (d) Left lateral profile showing a comparatively normal side; (e) Intraoral frontal view showing dental crowding with midline shift; (f) Occlusal view of mandibular arch showing moderate anterior crowding and arch asymmetry.

Routine complete blood count and biochemical analysis of renal and hepatic function were within normal limits, except for a slight elevation in erythrocyte sedimentation rate (ESR) and the presence of normocytic normochromic anemia. Antinuclear antibody (ANA) titer was 1:160 showing a speckled pattern with cytoplasmic positivity. Antineutrophil cytoplasmic antibody (ANCA) was 1:32, exhibiting a perinuclear pattern (p-ANCA) on indirect immunofluorescence. Immunoblot ANA profile demonstrated weak positivity for anti-RNP and anti-SSA antibodies, while tests for double-stranded DNA and anti-Scl-70 were negative.

Radiographic examination was conducted in the form of a Non-Contrast Computed Tomography (NCCT) scan of the head and neck region and an orthopantomogram (OPG) of the maxillary and mandibular region. The Sagittal view of NCCT head revealing hypoplastic sinuses, hypoplastic mandible, deviated nasal septum, with an abnormal Rt eye ball while no abnormality was detected in the brain, and

the OPG reveals a shorter and hypoplastic right mandibular ramus, with underdeveloped body and angle region. The right condylar head appears malformed along with a less prominent right coronoid process [Figure 2]. A midline shift is also noted, which is clinically visible as well. Histopathologic examination of a punch biopsy from the affected facial skin revealed marked dermal fibrosis with thickened collagen bundles. There was prominent perivascular and periadnexal lymphocytic infiltration extending into the reticular dermis. Epidermal atrophy was observed with loss of rete ridges, and adnexal structures showed signs of atrophy. No evidence of vasculitis or granuloma formation was seen.

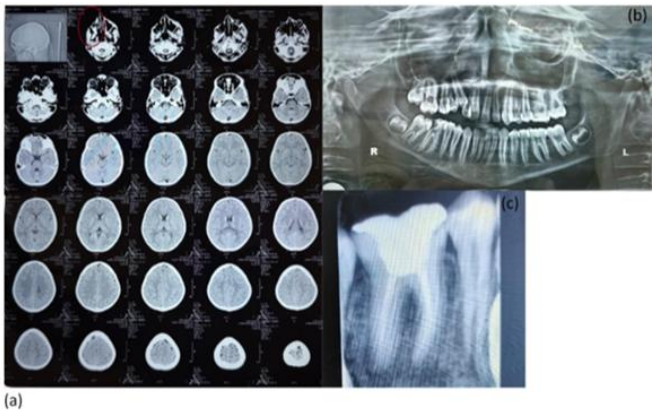

Figure 2: a) Sagittal view of NCCT head revealing hypoplastic sinuses, hypoplastic mandible, deviated nasal septum, with an abnormal Rt eye ball while no abnormality was detected in the brain; b) Orthopantomogram (OPG) which reveal hypoplasia of right mandibular angle and body region; (c) Intra-oral periapical radiograph showing obturated right mandibular molar with a seated Endocrown

As part of the dermatological management, topical treatment with 0.1% tacrolimus ointment was initiated to reduce inflammation and progression of localized scleroderma. Tacrolimus, a calcineurin inhibitor, is commonly used in pediatric patients for immune-modulated skin conditions due to its steroid-sparing benefits and efficacy in stabilizing disease progression.

On intraoral examination, the lower right mandibular molar region was tender on percussion. Clinical and radiographic evaluation confirmed the diagnosis of irreversible pulpitis in the permanent first molar (tooth 46). In view of the diagnosis, root canal treatment was initiated under local anesthesia using a standard aseptic protocol. Biomechanical preparation was performed using rotary NiTi instruments, followed by irrigation with sodium hypochlorite and saline. The canals were dried and obturated using gutta-percha and resin-based sealer. Given the tooth's structural loss and the need for functional and esthetic restoration, the patient was planned for a 3D-printed endocrown. An intraoral scan of the mandibular arch was obtained and it was used to digitally design and fabricate a custom-fitted zirconia endocrown, which was subsequently luted using dual-cure resin cement [Figure 3].

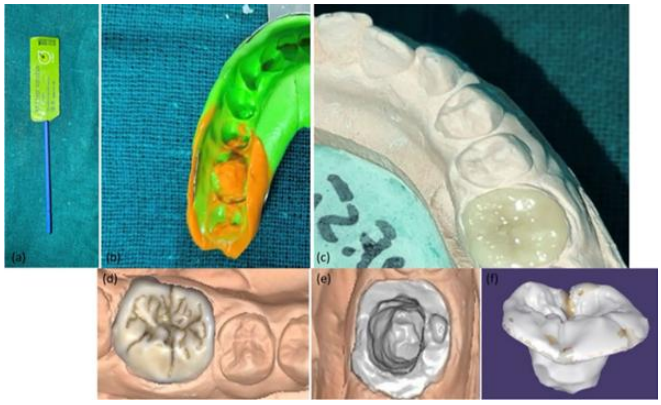

Figure 3: (a) Fluoride Varnish; (b)Elastomeric impression of crown preparation; (c) Digital design and fabrication of an Endocrown using CAD/CAM; (d) A computer-designed and milled Endocrown seated on a dental cast; (e) Final version of the endocrown created through CAD/CAM.

Additionally, topical fluoride varnish (5% sodium fluoride) was applied to the patient's teeth as a preventive measure. Given the heightened caries risk associated with facial asymmetry, fluoride varnish provides prolonged contact with the enamel surface, enhancing remineralization and reducing the risk of future demineralization. This intervention was especially important in the context of the patient's recent endodontic treatment.

3.Discussion

Juvenile scleroderma is the third most common rheumatic disease in children, following juvenile idiopathic arthritis and systemic lupus erythematosus (SLE). It primarily occurs in two forms: juvenile localized scleroderma (JLS) and juvenile systemic sclerosis (JSSc). Juvenile localized scleroderma (JLS) comprises a group of conditions primarily affecting the skin and subdermal tissues, with internal organ involvement being rare.[7] An epidemiological study from the United Kingdom reported an incidence of 3.4 cases per million children annually, with the linear subtype being the most prevalent.[8] The female-to-male ratio is 2.4:1, and the average age of onset is around 7.3 years, although cases can present as early as birth, as seen in congenital localized scleroderma. At such an early age, the condition is often misdiagnosed as a skin infection, nevus, or salmon patch, leading to significant delays in diagnosis.[7]

Localized scleroderma is marked by thickening and hardening of the skin and underlying tissues due to excessive collagen build-up. It includes a range of clinical forms, from superficial, cosmetically concerning morphea plaques to more severe variants such as linear scleroderma and eosinophilic fasciitis, which can result in significant morbidity. Unlike systemic sclerosis, localized scleroderma does not typically involve features like sclerodactyly, Raynaud's phenomenon, or internal organ involvement.[9]

The different forms of juvenile localized scleroderma (JLS) are classified based on the distribution and

characteristics of skin lesions, along with other associated features. The most commonly used classification includes five subtypes: circumscribed morphea (CM), linear scleroderma, generalized morphea, pansclerotic morphea, and a mixed subtype that presents with a combination of two or more of the aforementioned forms.[5]

1. Circumscribed morphea typically presents as oval or round areas of skin hardening, with a central waxy, ivory-colored plaque surrounded by a violaceous border. The superficial form is limited to the dermis, while the deep form affects the entire skin thickness and subcutaneous tissue, resulting in a thickened, tight, and immobile lesion. These lesions most commonly appear on the trunk, though they can also affect the extremities or face.[7]
2. Generalized morphea involves four or more individual lesions, each larger than 3 cm, which merge to cover at least two of seven anatomical regions: head/neck, right or left upper extremity, right or left lower extremity, anterior trunk, and posterior trunk. It may also occur in either superficial or deep forms.[7]
3. Linear scleroderma is the most frequent subtype in pediatric patients. It is characterized by one or more linear streaks that commonly affect the limbs and trunk. These streaks gradually become more hardened and may extend from the skin through subcutaneous tissue and muscle, sometimes even involving the underlying bone. Facial or scalp involvement occurs in the en coup de sabre (ECDS) variant, named for its resemblance to a sword strike indentation. Parry-Romberg syndrome (PRS), which presents as progressive hemifacial atrophy beneath the forehead with minimal or no superficial skin involvement, is considered a severe form of ECDS. Currently, both conditions are grouped under the broader term “linear scleroderma of the face”.[5,10]
4. Pansclerotic morphea is a very rare but severe form of the disease, marked by widespread, full-thickness skin involvement affecting the trunk, limbs, face, and scalp, while typically sparing the fingertips and toes. This condition is highly debilitating and can result in serious limb deformities, and in extreme cases, autoamputation due to fibrotic constriction and deep vascular occlusion. Unlike systemic sclerosis (SSc), pansclerotic morphea involves extensive skin changes without internal organ involvement, which helps in distinguishing between the two. Chronic ulcers are a common complication of this subtype, and studies have highlighted the potential risk of these ulcers progressing to squamous cell carcinoma.[7]

“Blaschko’s lines” describe a rare and poorly understood anatomical pattern first identified in 1901 by German dermatologist Alfred Blaschko.[11] These lines are typically invisible under normal circumstances but become apparent in certain inherited or acquired skin and mucosal conditions, where they appear as distinctive stripes. They are widely believed to reflect patterns of genetic mosaicism.[4] Several case reports have noted that lesions in linear morphea tend to follow Blaschko’s lines, suggesting a possible link to embryological development.[3] Soma and Fujimoto further proposed that the en coup de sabre variant of scleroderma also aligns with these lines.[12]

Managing and treating juvenile localized scleroderma can be difficult, as no definitive cure currently exists. Therapeutic approaches primarily aim to reduce inflammation and control collagen abnormalities.[4] Children with morphea also require specialized dental care, including preventive measures and regular monitoring of craniofacial growth and development. The overall prognosis for morphea is generally good, with minimal risk of systemic complications and a normal life expectancy.

This case underscores the importance of early dermatological and dental assessment in children presenting with facial asymmetry, potentially improving outcomes through timely, targeted interventions.

4.Conclusion

Juvenile localized scleroderma, particularly the linear en coup de sabre variant, can lead to significant esthetic, functional, and dental complications if not diagnosed and managed early. The condition often mimics other dermatologic presentations, causing diagnostic delays. Management requires a multidisciplinary approach involving dermatologists and dental specialists. Topical immunomodulators like tacrolimus have shown effectiveness in controlling disease progression. Dental interventions should include both curative and preventive strategies, especially in the presence of craniofacial abnormalities. With appropriate treatment, prognosis remains favorable despite the potential for long-term morbidity.

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