Case Report

Kindler Syndrome with Bleeding Gums and Bleeding Per Rectum: A Case Report

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Abstract

Kindler syndrome is a rare genetic skin disorder caused by a mutation in chromosome 20's short arm that leads to the loss of kindlin-1 function. Kindlin-1 plays an important role in keratinocyte adhesion. The syndrome is characterized by skin fragility and various skin problems such as photosensitivity, skin discoloration, inflammation of the mucous membrane, bleeding gums, blistering, skin atrophy, and poor dentition. We present a unique case of a 12-year-old boy with Kindler syndrome who had bleeding gums. It requires a multidisciplinary approach and genetic counseling. Due to genetic heterogeneity, prenatal diagnosis is challenging.

Keywords: Genodermatosis, Epidermolysis bullosa, Kindler syndrome, Photosensitivity, skin atrophy, poikiloderma.

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Introduction

A 14-year-old male patient presented to the dermatology outpatient department with a history of photosensitivity, bleeding per rectum, and gum bleeding for one month. The patient was born to nonconsanguineous parents with four siblings, and his sister died of hemorrhage due to colitis and Kindler syndrome.

On cutaneous examination, fragile skin, poikiloderma of skin, dorsum of hands, and dorsum of feet skin wrinkling, oral mucosal fragility, bleeding on slight injury, mild palmoplantar keratoderma, and loss of dermatographism were observed. The patient was managed with sunblock, moisturizers, fusidic acid cream, and gentle teeth-brushing techniques. On general physical examination, the pulse was 78 per minute, respiratory rate was 16 per minute, afebrile, and blood pressure was 110/75 mm of Hg. Systemic examination was unremarkable.

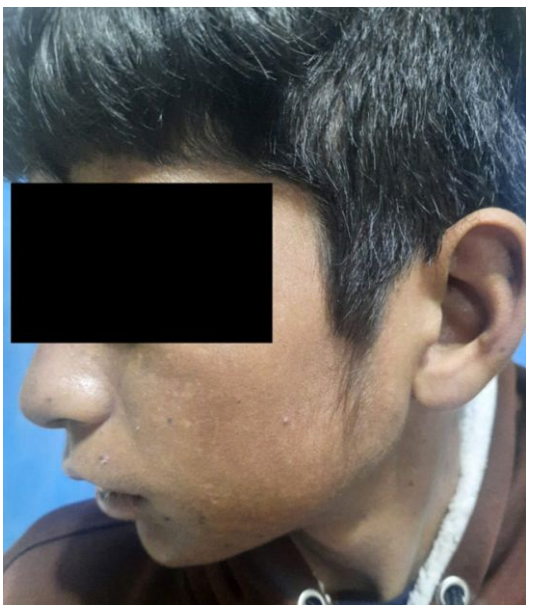

Figure 1: *Photosensitivity and hyperpigmentation of the face*

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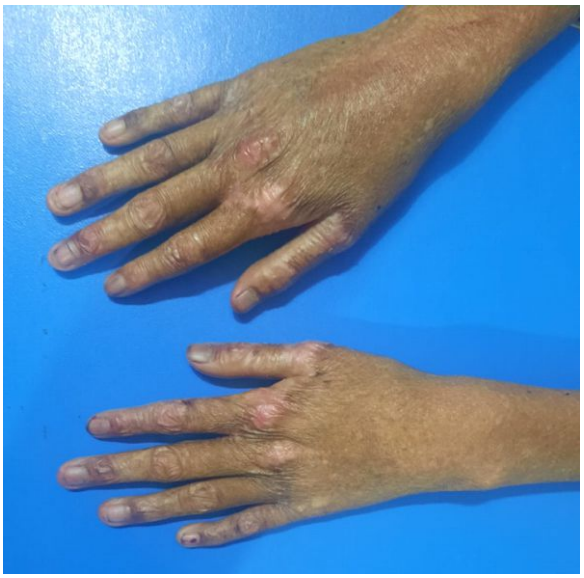

Figure 2: *Hypopigmentation, hyperpigmentation, atrophy and telangiectasia, poikiloderma*

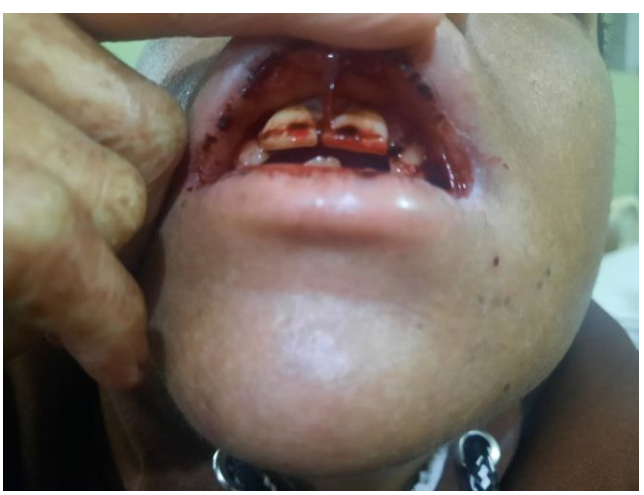

Figure3: *Oral cavity showing gum bleeding*

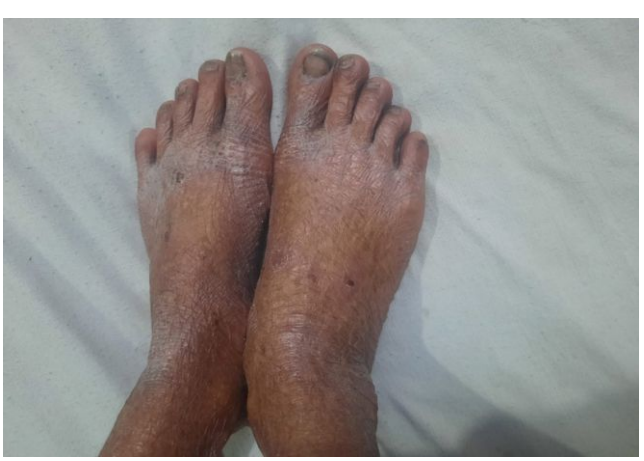

Figure 4: *Skin of feet showing xerosis and cigarette paper wrinkling*

Figure 5: *Anal cavity examination showing mucosal ulceration*

Differential diagnoses included

Bloom's syndrome, Cockayne Syndrome, Xeroderma Pigmentosum, Dyskeratosis Congenita, and Rothmund-Thombson syndrome.

Introduction

The first case of Kindler's syndrome was reported in 1954.[1] Since then, more than 250 to 400 cases have been reported.[1,2,3]

Besides skin changes, genitourinary, gastrointestinal, and oral mucosa are also affected. FERMT1 gene mutation is responsible for most of the cases. Studies suggest FERMT1 gene mutations are absent in 30% of cases.[1] The histopathological features of keratinocytes are attenuated due to Kindlin 1 deficiency, leading to premature aging and fragility of keratinocytes. Dermal and intraepidermal cleavage is seen on ultra-structural skin examination.[1] Other clinical findings include pseudo syndactyly, hyperkeratosis of palms and soles, and periodontitis in the oral cavity.[2] Mucosal involvement may range from minor ulcerations to stenosis in the esophagus, anus, urogenital region, cornea, and conjunctiva.[2] Associated malignancies may be squamous cell carcinoma of the skin and larynx, or transitional cell carcinoma of the bladder.[2] Management may be multidisciplinary and involve a dermatologist, ophthalmologist, urologist, gastroenterologist, dentist, nutritionist, or psychologist.[2] Around 55 FERMT 1 mutation variants including deletion, insertion, missense, and splice site mutation have been detected so far.[3]

Discussion

The prevalence may occur with a frequency of 1 case per 50,000 births annually, whereas severe cases could be diagnosed in 1 case per 500,000 births annually.[4] Both autosomal recessive and autosomal dominant variants have been identified.[4] Kindler syndrome may involve the skin and mucous membranes with radiological changes evidence.[4] The genetic mutation involved may encode keratin, desmosomes, or hemidesmosomes.[4] Genetic testing of the fetus in high-risk families through chronic villus sampling at twelve weeks of gestation or amniotic fluid samples at sixteen weeks of gestation allows the prediction of the disease.[4] Good wound care prevents complications of skin wounds like sepsis.[4] Moisturizers are helpful together with protecting fragile skin from injury and sun damage.[4] Topical dressings, lipid colloid dressings, and hydrogel sheets may be used for skin wound dressings.[4] Pain and inflammation can be controlled with effective oral analgesics.[4] In case of secondary infection, topical and oral antibiotics can be helpful.[4]

Unlike other forms of epidermolysis bullosa, in Kindler's syndrome, there is a reduction in the number of blisters forming with the progression of age.[5] Photosensitivity occurs at variable degrees of severity among the patients. However, skin atrophy manifests before the age of five years mostly involving the back of hands and feet.[5] Skin atrophy becomes widespread during adolescence.[5] Kindler syndrome severe colitis and constipation may develop iron deficiency anemia.[5] Other systemic manifestations are nail dystrophy, leukokeratosis of lips, pseudoainhum, dental caries, hypohidrosis, and palmoplantar keratoderma.[5]

Skin biopsy and histopathology in Kindler syndrome are inconclusive: it may show hyperkeratosis, skin atrophy, attenuated and flattened epidermis, and loss of elastic fibers in the papillary dermis with mild infiltration of macrophages and lymphocytes. The dermis contains dilated vessels, dermal edema, and fibrosis.[5]

Radiological screening shows delayed epiphyseal fusion.[6] Kindler's syndrome may present with short stature, recurrent infection, and frequency of hematological malignancies.[6,7] The chances of squamous cell carcinoma can be developed in ten percent of patients after the age of forty-five years.[7] Loss of fingerprints or absent dermatographia has been described with Kindler syndrome.[8] Patients with Kindler syndrome have a normal lifespan but are associated with comorbidities. Diagnostic criteria include five major and two minor clinical diagnostic criteria; major criteria include acral blistering in infancy and childhood, progressive poikiloderma, skin atrophy, photosensitivity, gingival fragility, and swelling. The minor proposed criteria are syndactyly and mucosal involvement (anal, esophageal, urethral, and laryngeal stenosis). The presence of four major criteria for the diagnosis is certain. The presence of three major and two minor criteria makes the clinical diagnosis probable, and the diagnosis is considered likely if two major and two minor criteria are present.[9] Sun protection and the use of sunblock are the mainstay in managing photosensitivity. It also delays the onset of poikiloderma. Management of patients is symptomatic and prevention of trauma, wound care, good nutritional and psychological support.[7,8,9]

Conclusion

We report this case of Kindler syndrome as it is rare and its complications like colitis need urgent management and consultation with a gastroenterologist needs multidisciplinary management involving a dermatologist, gastroenterologist, dentist, nutritionist, and psychologist for the patient's overall well-being. The nutritionist will assess the nutritional deficiencies of this patient. The psychologist will provide parental counseling and overall psychological support for the emotional well-being of the patient.

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Authors Contribution

HS: Conceptualization of Project
HS: Data Collection
MJ, SR: Literature Search
MJ, SR: Statistical Analysis
MJ, SR: Drafting, Revision
HS, NAA: Writing of Manuscript