CASE REPORT

Acrodermatitis Enteropathica

Chinmai Sanjay Shinde*, Vikrant Jadhav, Anil Gugle, Paresh Agiwal, Th. Meghna Devi, Shivani Shinde

Abstract

Acrodermatitis enteropathica is an uncommon genetic condition inherited in an autosomal recessive manner. It results from impaired Zn-absorption, causing systemic deficiency of this essential mineral and giving rise to diverse dermatological and systemic symptoms. We report a case of a 35-year-old female presented with a longstanding history of vesicular bullous lesions on hands and feet, oral lesions, diarrhoea, and hair loss. Zinc supplementation was initiated which led to symptom improvement. Early recognition and management of zinc deficiency is crucial in preventing long-term complications. Zinc supplementation and dietary modifications effectively treated acrodermatitis enteropathica in this case.

Keywords: Acrodermatitis Enteropathica, Zinc deficiency

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Introduction

Acrodermatitis enteropathica (AE) is a genetic disorder inherited in an autosomal recessive pattern, leading from zinc deficiency. This condition arises from impaired absorption of dietary zinc in the duodenum and jejunum.[1] Zinc serves crucial roles as a co-enzyme in various metalloenzymes, such as alkaline phosphatase, and as a structural component in gene regulatory proteins necessary for cellular functions like tyrosine kinase binding to T-cell receptors. It also plays a role in gene expression regulation.

Severe zinc deficiency manifests with diverse signs and symptoms, including growth retardation, compromised immune function, and various skin or gastrointestinal lesions.[2]

Case Representation

Clinical Presentation: A 35-year-old female presented to the Outpatient Department (OPD) with chief complaints of dark lesions on her hands and feet dating back to childhood. Approximately 25 years ago, she first experienced fluid-filled lesions on both hands and feet. The lesions initially appeared on her hands followed by her feet. These lesions would spontaneously rupture within 2-3 days, releasing watery fluid, and subsequently heal with dark pigmentation within 10 days. They were associated with pain, burning sensations, and itching. The patient has had multiple episodes of such lesions since childhood.

Additionally, the patient complained of flat reddish lesions on her upper lip, lower lip, and angles of the lips. These lesions would darken over time, and she has experienced similar lesions on and off since childhood. She also developed dark pigmentation on her tongue. The patient also reports gradual, progressive hair loss over 20 years. She also experienced 2-3 episodes of diarrhea per month over the past year. In the last six months, she has noticed dark oral lesions on her tongue and buccal mucosa, associated with pain and a burning sensation.

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Dermatological Examination

On examination, multiple well-defined hyperpigmented healed erosions covered with crust and borders showing exfoliation, measuring approximately 4 × 8 cm, were present on the dorsum of feet. There were multiple hyperpigmented macules on the tongue and buccal mucosa. Diffuse thinning of hair was noted. Hyperpigmented macules were also observed in the cubital fossa (Figure 1).

Autoimmune bullous disorders, Atopic dermatitis, and zinc deficiency were considered in differential diagnosis.

Investigations

We investigated for complete blood count, LFT, RFT, Serum Zinc levels, Serum B12 levels, skin punch biopsy. The serum zinc levels (32 mcg/dL), alkaline phosphatase and albumin levels were low. Histopathological examination of a skin biopsy showed hyperkeratotic stratified squamous epithelial layers with an increased granular cell layer, along with mild to moderate inflammatory infiltrate and dermal edema in the underlying dermis. There were no remarkable findings in the adnexal structures (Figure 2).

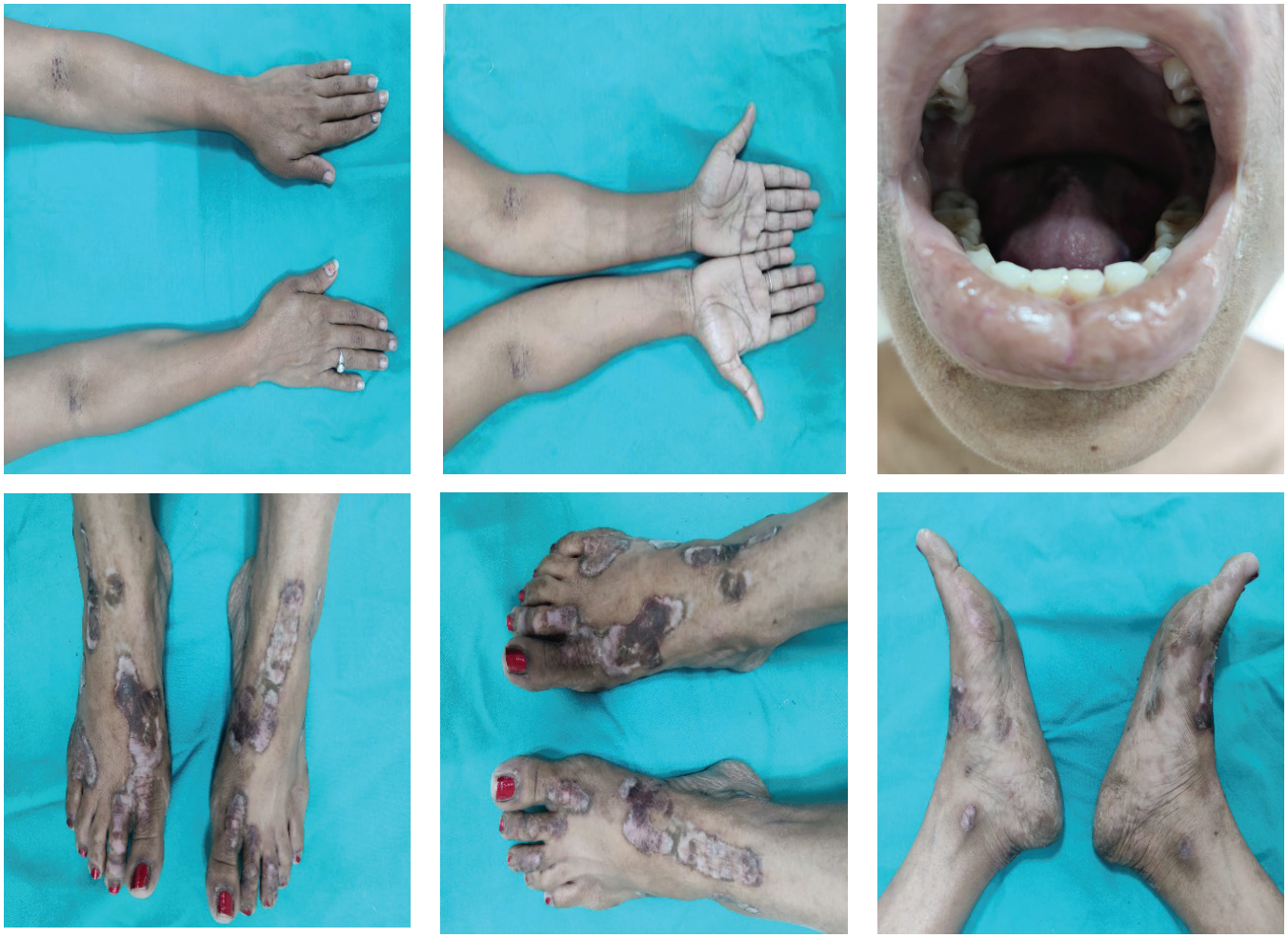

Figure 1: Dermatological Examination.

Treatment: Dose of elemental zinc, 2 mg/kg/day, [Tablet Zinc Acetate (elemental zinc 50 mg)] BD for one month showed improvement. Then the dose was reduced to 1-mg/kg/day [Tablet Zinc Acetate (elemental zinc 50 mg)] OD after symptom control. The patient was also advised to increase her intake of food products rich in zinc like seafood, cashew and other nuts, whole grains, cereals, and dairy products.

Outcomes: Skin lesions and dermatitis typically began to resolve within a few days after zinc supplementation was initiated.

Discussion

Acrodermatitis enteropathica (AE) is a rare autosomal recessive disorder. It is characterized by severe zinc deficiency, which arises from impaired absorption of dietary zinc in the duodenum and jejunum.[1] Zinc is essential as a cofactor for various metalloenzymes and is critical for cellular functions, including gene expression regulation and immune response.

AE typically manifests in early childhood following the cessation of breastfeeding. Symptoms vary by age group; infants may exhibit diarrhea, mood changes, anorexia,

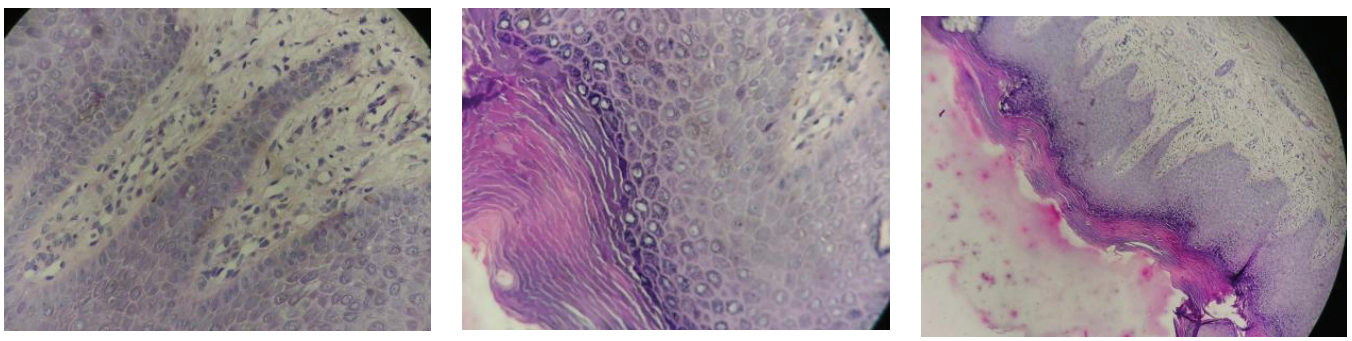

Figure 2: Histopathological examination of a skin biopsy.

and neurological disturbances. School-aged children and toddlers often experience growth retardation, alopecia, weight loss, and recurrent infections. Spontaneous remission can occur during adolescence.[3]

Diagnosing AE involves challenges; laboratory tests measuring zinc levels in serum, urine, or hair lack specificity and sensitivity. Zinc absorption tests are complex, while genetic testing for mutations in the SLC39A4 gene on chromosome 8q24 provides definitive diagnosis but is not universally available.[2] Clinicians often rely on the immediate response to therapeutic zinc supplementation (3–30 mmol/kg body weight) to guide diagnosis and treatment decisions.

Zinc deficiency dermatitis presents as either "acrodermatitis enteropathica" or "acquired acrodermatitis enteropathica," depending on its cause. Both conditions are managed similarly but differ in etiology.[4] Acrodermatitis enteropathica results from genetic mutations affecting zinc transporters, typically appearing in infancy. In contrast, acquired acrodermatitis enteropathica arises from dietary zinc deficiency and can also affect premature infants with higher zinc demands.[5]

In our case, treatment involved zinc supplementation leading to rapid healing of skin lesions. Long-term management includes monitoring zinc levels and adjusting supplementation as needed, ensuring favourable outcomes with sustained intervention.

Conclusion

This case highlights a multifaceted presentation of zinc deficiency, manifesting as a distinct dermatological, oral, and gastrointestinal symptom complex in a 35-year-old female. Timely initiation of zinc supplementation led to substantial clinical improvement. The importance of identifying and addressing nutritional deficiencies in such cases is underscored. Long-term management involving dietary adjustments and ongoing zinc therapy effectively resolved symptoms, underscoring the significance of early intervention in mitigating complications linked to zinc deficiency.

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