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Acrodermatitis Enteropathica in an Infant with Normal Serum Zinc Level: A Case Report

H. El Halla[1*], M. Chaouche[2], S. Mhaimer[1], I. Zouine[1], N. Kayouh[1], H. Lahmidi[1], C. Aassali[1], S. Chhiti[2], R. Chakiri[1]

[1]Department of Dermatology, Faculty of Medicine and Pharmacy, Ibn Zohr University, Agadir, Morocco
[2]Faculty of Medicine and Pharmacy, Ibn Zohr University, Agadir, Morocco

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***Corresponding author:** H. El Halla
Department of Dermatology, Faculty of Medicine and Pharmacy, Ibn Zohr University, Agadir, Morocco

Abstract **Case Report**

Acrodermatitis enteropathica is a rare dermatological disorder related to impaired zinc metabolism, characterized by a typical clinical presentation, although biological findings may occasionally be non-contributory. We report a case of a 9-month-old infant, exclusively breastfed, presenting with progressive erythematous and crusted cutaneous lesions associated with severe diarrhea and a systemic inflammatory response. Clinical examination revealed extensive periorificial dermatitis, cicatricial alopecia, bullous and purpuric lesions, and bilateral ectropion. The diagnosis of acrodermatitis enteropathica was suspected based on the characteristic clinical triad, despite normal serum zinc level. The diagnosis was supported by histopathological findings and, most importantly, by a rapid clinical response to zinc supplementation. The clinical course was rapidly favorable, with early improvement of cutaneous lesions and resolution of the inflammatory syndrome. This case highlights that normal serum zinc level do not exclude the diagnosis of acrodermatitis enteropathica and emphasizes the central role of therapeutic zinc supplementation as both a diagnostic and therapeutic tool.

Keywords: Acrodermatitis enteropathica, Zinc deficiency, Periorificial dermatitis, Alopecia, Diarrhea, Infant.

INTRODUCTION

Zinc is an essential trace element in the human body, playing a crucial role in numerous physiological processes. It acts as a cofactor for a wide range of enzymes, thereby participating in various metabolic reactions. Beyond its enzymatic function, zinc also serves a key structural role by contributing to the stabilization of cellular structures, particularly biological membranes and nuclear proteins such as histones. Furthermore, it is required for the activity of nucleic acid polymerases as well as various intracellular enzymes involved in protein biosynthesis. These properties confer upon zinc a central role in mechanisms of cellular proliferation, differentiation, and tissue growth.[1] Consequently, zinc is essential for the proper functioning of multiple organ systems, particularly the gastrointestinal, nervous, and cutaneous systems.[1]

Acrodermatitis enteropathica is a rare inherited disorder with autosomal recessive transmission, caused by zinc deficiency. It represents the prototypical clinical expression of zinc deficiency, arising from either impaired intestinal absorption or insufficient dietary intake. Clinically, it is classically defined by a triad of periorificial dermatitis, alopecia, and diarrhea, often accompanied by additional ocular, gastrointestinal, and mucocutaneous manifestations with variable presentation and frequency. [2] Early diagnosis is crucial, as zinc supplementation leads to rapid clinical improvement and helps prevent the development of complications.

We report the case of a 9-month-old full-term infant, exclusively breastfed, in whom a diagnosis of acrodermatitis enteropathica was established despite normal serum zinc levels, with marked clinical improvement following zinc supplementation.

CASE REPORT

A 9-month-old infant, born at term to non-consanguineous parents, exclusively breastfed, with no family history of similar lesions, was admitted for erythematous crusted lesions evolving over one month. The lesions were initially localized to the diaper area and neck, with progressive extension, and were associated with pruritus and profuse diarrhea (>8 stools/day). One

week prior to admission, the clinical condition deteriorated, with the development of tense blisters associated with a fever of up to 39°C.

Dermatological examination revealed extensive erythematous, crusted, and scaly plaques with a periorificial distribution (perioral, periorbital, and perineal) and involvement of the skin folds, along with scattered purpuric lesions. The remainder of the physical examination showed a tense blister on the right hand with a negative Nikolsky sign. Scarring alopecia was noted. Ophthalmologic examination revealed bilateral ectropion.

Given the triad of periorificial dermatitis, alopecia, and diarrhea, a diagnosis of acrodermatitis enteropathica was suspected. Zinc supplementation was initiated at a dose of 5 mg/kg/day, and a skin biopsy was performed. Laboratory investigations were overall unremarkable (serum zinc: 11.7 µmol/L; vitamin B12: 372 pg/mL), except for an elevated C-reactive protein level of 157.2 mg/L, while alkaline phosphatase levels were within normal limits.

Histopathological examination supported the diagnosis, showing parakeratosis, a reduced granular layer, suprabasal vacuolization, and a mild perivascular inflammatory infiltrate.

A rapid clinical improvement was observed within 3 days, along with a decrease in C-reactive protein to 10.7 mg/L after 6 days.

DISCUSSION

Acrodermatitis enteropathica (AE) is a rare genetic disorder caused by impaired intestinal absorption of zinc. Its prevalence is estimated to range from 1 to 9 cases per 1,000,000 inhabitants, with a global incidence of approximately 1 case per 500,000 live births. It occurs across all populations, with no predilection for sex or ethnic background.[3] The etiology may be either genetic or acquired. The acquired form can be associated with various conditions, including malabsorption syndromes, inflammatory bowel diseases, chronic pancreatitis, short bowel syndrome, celiac disease, cystic fibrosis, and prolonged parenteral nutrition. Patients with malignancies, burns, or infections may develop zinc deficiency due to increased metabolic demands. Certain medications, such as penicillins, diuretics, valproate, antimetabolites, and iron supplementation, may also contribute to deficiency of this micronutrient. [4] In a review of the literature by Wanlin Cui *et al.,*, the two main causes of zinc deficiency were insufficient dietary intake and impaired intestinal absorption [5]. In genetic forms, the disease typically manifests during the first months of life.

Initial clinical manifestations are often dominated by nonspecific cutaneous lesions. Early lesions typically consist of erythematous plaques, xerosis, and eczematous changes, which may delay diagnosis due to their lack of specificity.

Cutaneous involvement classically begins in acral and periorificial areas, with a predilection for the face, scalp, and genital region, as observed in our patient. The course is usually rapid, with progressive extension and worsening of dermatological lesions, which may acquire a more pronounced inflammatory appearance.

During disease progression, additional signs may emerge, including paronychia, palmar involvement, and a wrinkled or puckered appearance of the digital pulps, reflecting epidermal damage related to zinc deficiency.

Mucosal involvement is also common and constitutes an integral part of the clinical presentation. It mainly manifests as glossitis, stomatitis, and angular cheilitis, highlighting the vulnerability of rapidly renewing tissues to zinc deficiency.

Overall, this polymorphic clinical presentation, combined with its rapid progression, underscores the importance of early diagnosis in order to enable appropriate zinc supplementation and prevent both systemic and cutaneous complications.[6]

Diarrhea is a major feature of acrodermatitis enteropathica, and its presence plays a crucial role in establishing the diagnosis. It constitutes part of the characteristic clinical triad, which includes acral and periorificial dermatitis, diarrhea, and alopecia.[7] The latter is often an early and revealing feature, as illustrated by our patient, who presented with chronic, diffuse non-scarring alopecia. However, the complete clinical triad is observed in only 20% of patients.[8]

The diagnosis is primarily clinical and may be supported by serum zinc measurement; however, normal zinc levels do not exclude the diagnosis. In this context, decreased levels of zinc-dependent enzymes, such as alkaline phosphatase, may provide supportive evidence for the diagnosis of Acrodermatitis Enteropathica.[2]

Differential diagnoses may include congenital impetigo, psoriasis, candidiasis, seborrheic dermatitis, Stevens–Johnson syndrome, and atopic dermatitis.

Histopathological examination is nonspecific, it typically reveals parakeratosis, a reduced granular layer, suprabasal vacuolization, and a mild perivascular inflammatory infiltrate.[2]

The gold standard of management is zinc supplementation, which may also serve as a therapeutic test. The recommended doses of zinc vary across the literature, with some authors suggesting 1–2 mg/kg/day and others up to 5 mg/kg/day. In our patient, zinc sulfate was administered at a dose of 5 mg/kg/day, resulting in

marked clinical improvement within 3 days of treatment. In severe cases, intravenous administration of zinc chloride at a dose of 10 to 20 mg is recommended.[2]

Clinical improvement is generally rapid following treatment initiation. Digestive symptoms, particularly diarrhea, typically resolve within 24 hours. Cutaneous signs begin to improve within the first 24 hours, while complete healing of severe skin lesions is usually achieved after approximately one week of therapy. [9]

Lifelong zinc supplementation, combined with a zinc-rich diet, is required to prevent relapse.[10] However, some authors suggest continuing zinc supplementation only until the introduction of complementary feeding.

CONCLUSION

This reported case highlights that the diagnosis of acrodermatitis enteropathica should not be excluded in the presence of normal serum zinc levels, particularly when the clinical presentation is suggestive. Laboratory parameters may be misleading or influenced by prior supplementation or by the dynamic course of the disease. Therefore, the diagnosis remains primarily clinical.

Early therapeutic testing with zinc supplementation has both diagnostic and therapeutic value and is typically associated with a rapid and often dramatic clinical response.

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