CASE REPORT

Diagnosis of acrodermatitis enteropathica in resource limited settings

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SUMMARY

Acrodermatitis enteropathica (AE) is a rare inherited zinc deficiency that usually manifests in infancy within days in cases of bottlefed infants and days to weeks after weaning in breastfed infants. It is characterised by diarrhoea, dermatitis, alopecia and systemic symptoms. We report a case of acquired nutritional AE in a 6-month-old female infant who had diarrhoeal episodes and the characteristic dermatitis lesions in the acral and anogenital regions. She responded dramatically to oral zinc supplementation.

BACKGROUND

The diagnosis of acrodermatitis enteropathica (AE) is based on the characteristic morphology and distribution of the skin lesions. The presence of eczematous pink scaly plaques over the acral extremities and anogenital areas with the background history of diarrhoea is suggestive of AE. A serum zinc level of 50 µg/dL confirms the diagnosis of AE. The dramatic therapeutic response to oral zinc supplementation

in this case is pathognomonic of AE. The diarrhoeal episodes stopped, and the skin lesions improved within 48 hours and became normal within a week. This therapeutic response could be considered as a confirmatory diagnostic test particularly in resource poor settings where the facility for estimation of zinc is not freely available.

Case presentation

A 6-month-old female infant was brought to our skin outpatient department (OPD) for evaluation of diarrhoea and skin lesions. She had 4–5 episodes of watery stools daily for the past 5 months, which did not result in dehydration or hospitalisation. Her mother noticed flaky skin lesions in the buttocks and acral parts for the past 3 months that did not improve with topical medications. She did not have any other cutaneous and/or systemic complaints. She was exclusively breastfed and born to non-consanguineous parents. The developmental milestones were normal. Her elder sibling had skin rash that had lasted for about 2 months during her infancy,

Figure 1 (A and B) Dry erythematous and eczematous plaques in the perineum, knees, feet and gluteal region. (C and D) Complete improvement of skin lesions after 1 week of oral zinc supplementation.

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Figure 2 (A and B) Completely normal skin except for pigmentary changes, after 6 weeks of oral zinc supplementation.

which improved after multivitamin supplementation. However, she did not have associated diarrhoea.

On physical examination, her height was 67.5 cm (above 50th centile) and weight was 6 kg (below 50th centile). There was no significant lymphadenopathy or organomegaly. Neurological and other systemic evaluation was normal. Cutaneous examination revealed sharply defined dry erythematous and eczematous plaques in the perineum, gluteal region, knees, hands and face (figure 1A,B). Hair and nails were normal. In view of acral and anogenital dermatitis with background history of diarrhoea, a diagnosis of zinc deficiency (AE) was considered.

OUTCOME AND FOLLOW-UP

The child was given oral elemental zinc at a dose of 3 mg/kg/day. There was abrupt cessation of diarrhoea and improvement in the skin lesions within 2 days. Skin lesions resolved completely (figure 1C,D) within a week. The child was reviewed after 6 weeks of zinc supplementation. The skin has become completely normal except for pigmentary changes (figure 2A,B). The parents were counselled regarding the continuation of the lowest effective maintenance therapy with regular monitoring of zinc level and close follow-up after weaning.

DISCUSSION

Zinc is an important trace element and has catalytic, structural and regulatory functions. It has a key role in the growth, tissue repair, wound healing, testicular hormone synthesis, immune response and keratinocyte metabolism.^{1 2} Zinc deficiency can develop in two forms: (1) inherited AE and (2) acquired nutritional zinc deficiency that results from poor nutrition, malabsorption states, malignancy, infection, total parenteral nutrition, renal disease and certain drugs.¹⁻³

AE is a rare inherited zinc deficiency that occurs worldwide. It is due to mutations in SLC39A4 gene that encodes a protein Zip4 zinc transporter, which are expressed in the enterocytes and help in the absorption of zinc. AE usually manifests in infancy within days in cases of bottlefed infants and days to weeks after weaning in breastfed infants. The clinical triad of AE includes acral and periorificial dermatitis, diarrhoea and alopecia; however, all the three features are seen only in 20% of the cases. Cutaneous lesions are often the initial manifestation and consist of eczematous pink scaly plaques, which may become vesiculobullous, pustular or psoriasiform. The sites typically affected are acral extremities and periorificial areas. In the absence of treatment, AE progresses and infants develop diarrhoea and alopecia. Features of advanced AE are failure to

Learning points

- ▶ Zinc deficiency occurs in two forms: (1) inherited acrodermatitis enteropathica (AE) and (2) acquired nutritional zinc deficiency.
- ▶ AE usually manifests in infancy within days in cases of bottlefed infants and days to weeks after weaning in breastfed infants.
- ▶ AE, like clinical presentation, also is seen in acquired nutritional zinc deficiency that may be seen even in breastfed infants due to low level of zinc in the breast milk or deficiency of zinc-binding ligand in breast milk.
- ▶ The dramatic therapeutic response could be considered as a confirmatory diagnostic test for zinc deficiency, particularly in resource poor settings where the facility for estimation of zinc is not freely available.

thrive, growth retardation, mental slowing, hypogeusia, delayed puberty, hypogonadism and several others.¹⁻³

Our case was an exclusively breastfed infant, and hence the deficiency is acquired nutritional rather than the rarer autosomal-recessive form of AE that typically develops after weaning off human breast milk. It has been hypothesised that it is due to the low level of zinc in the breast milk or deficiency of zinc-binding ligand in breast milk.^{4 5}

Diagnosis is usually based on the characteristic clinical morphology of the skin lesions. A serum zinc level of 50 µg/dL confirms the diagnosis of AE. A low serum alkaline phosphatase level is helpful.² The dramatic clinical improvement with zinc supplementation also confirms the diagnosis of zinc deficiency and this therapeutic response could be considered as a highly diagnostic feature particularly in resource poor settings where the facility for estimation of zinc is not readily available.

Infants with AE are treated with 3 mg/kg/day of elemental zinc.^{2 3} The parents should be counselled about the zinc deficiency, regular follow-up and the need of continuation of the lowest effective maintenance therapy with regular monitoring of zinc levels as the congenital form of AE requires lifelong zinc supplementation. However, in acquired nutritional zinc deficiency, there is gradual lessening of the need for continuous supplementation.

In conclusion, we report a case of acquired nutritional AE who showed an excellent and immediate therapeutic response. Such treatment response could be considered as a confirmatory test for AE in resource-limited settings.

Contributors TD and GS evaluated, assessed and managed the case. TD and GS wrote the manuscript, critically revised and approved it for submission.

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