Skin is a Mirror of Internal Disease: A Case of Acrodermatitis Enteropathica with Delayed Presentation

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ABSTRACT

Acrodermatitis enteropathica is a rare autosomal recessive disorder caused by a genetic mutation leading to zinc deficiency. Clinical manifestation includes skin lesions, diarrhea and alopecia. Here we report a case of five-year-old girl with alopecia, erythematous scaly crusted plaques over perianal, perineal areas, knees and ankle and toe nail dystrophy. With a clinical diagnosis of Acrodermatitisenteropathica, she was started on zinc therapy and her lesions subsided entirely after one month of treatment.

Keywords: Acrodermatitis enteropathica, Zinc deficiency

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CASE DESCRIPTION

A 5 year old, female child, born normally at full term to second degree consanguineous parents, presented with complaints of skin lesions and loss of hair since the age of 18 months. They visited many hospitals, treated with multiple topical steroids & antifungals. Lesions showed no response to any medications and new lesions continued to appears. Cutaneous examination revealed Bilaterally symmetrical erythematous scaly crusted plaques presented over perianal & perineal areas, dorsal aspects knees and ankle. Toe nails showed dystrophy. Scalp hair was dry and brittle with areas of alopecia. Weight and height of child was only 12.5 kg,98.5 cm respectively. Clinical diagnosis made as Acrodermatitis Enteropathica.

INVESTIGATIONS & TREATMENT

Alkaline phosphatase enzyme was 116 IU/L and Plasma Zinc level was low as 67mcg/dl. The patient was treated only with oral elemental zinc 3mg/kg/day. There was dramatic response to treatment in 7 days and skin lesions healed in one month, with regrowth of scalp hairs in two months.

DISCUSSION

We report a case of Acrodermatitis enteropathica affecting a child of second degree consanguineous parents. The diagnostic suspicion of zinc deficiency was initially supported by the topography of the lesions and no response to topicals. This was later confirmed by low serum zinc levels and immediate improvement with oral Zinc therapy.

Acrodermatitis Enteropathica (AE) is an inborn error in zinc metabolism, inherited as autosomal recessive

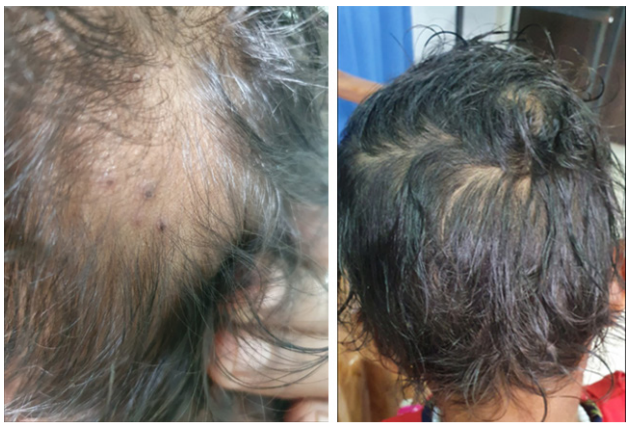

Figure 1. **Alopecia scalp. Scalp hair-Before and after treatment**

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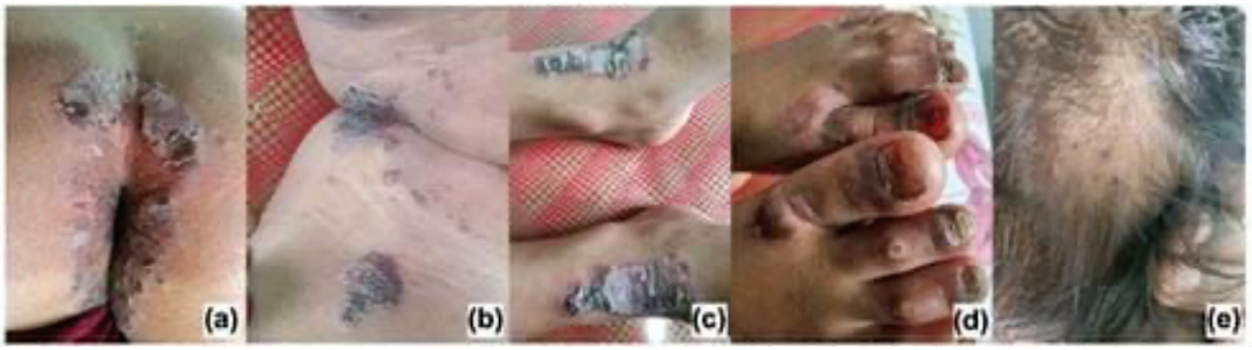

Figure 2 (a-e). **Clinical images at the time of Presentation**
Erythematous scaly crusted plaques over perineal, perianal, knees and ankle. Dystrophy of toe nails

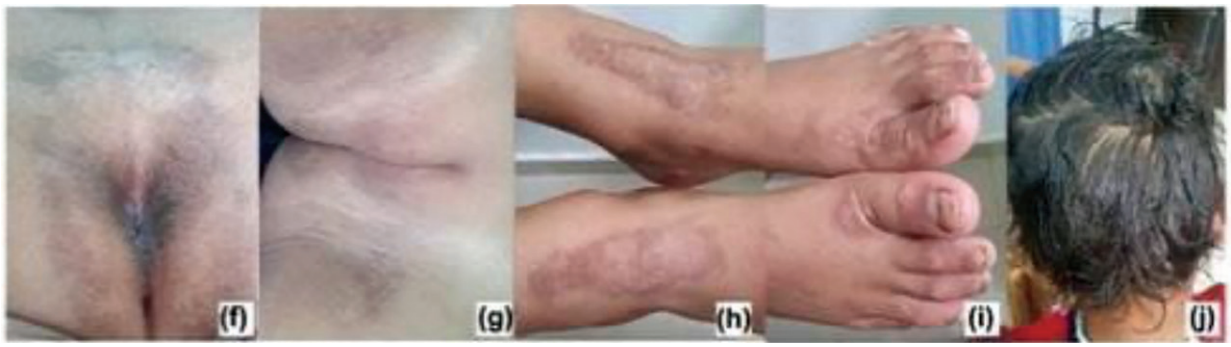

Figure 3 (f-j). **Clinical images after treatment**

disorder due to mutation in SLC39A4 gene encoding transmembrane zinc transporter protein **(Figure 1)**. The classic triad of AE include periorificial, intertriginous and acral dermatitis, alopecia and diarrhoea. Zinc is an essential trace element required for proper functioning of all cells, as it is the cofactor of numerous metal enzymes. Zinc deficiency can cause repeated infections, neurological disturbance and growth retardation **(Figure 2).**

CONCLUSION

When presented with symmetrical periorificial, anogenital and acral skin rash, with no improvement with topicals, physician should consider about Zinc deficiency Clinical images after treatment **(Figure 3)**. In some patients, even if the Zinc level is in normal range, if the skin lesions are characteristic, a trial of zinc supplementation should be considered. Zinc deficiency can be fatal, high index of suspicion is required to diagnose the condition, has to be investigated and treated early to prevent possible complications.

END NOTE

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Conflict of Interest: None declared

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