

Acrodermatitis Enteropathica in a 9 Month Old Infant

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A 9 month old female infant presented with a 4-week history of watery diarrhea, weight loss of 1.5 kg and irritability. She also had a 3-month history of a rash that did not respond to topical steroids or systemic antibiotics. The patient was the first child of consanguineous parents of Arab descent. She was born after an uneventful pregnancy and delivery and had an unremarkable perinatal period with breastfeeding until the age of 4 months.

Physical examination revealed multiple erythematous, thin, scaly papules and coalescing plaques symmetrically distributed in the perioral and perineal areas and on the cheeks, elbows and knees [Figure 1]. Laboratory tests were normal except

for a low serum zinc level (Zn 21, reference range 70–130 $\mu\text{mol/dl}$). The tentative diagnosis was acrodermatitis enteropathica. Treatment with oral zinc sulfate led to rapid healing of the skin lesions and alleviation of the diarrhea [Figure 2]. Genetic analysis showed the patient was homozygous for a SLC39A4 gene mutation. Her parents were heterozygous for the same mutation.

Acrodermatitis enteropathica is an autosomal recessive metabolic disorder caused by an inability to absorb zinc from the diet [1,2]. As found in our patient, the genetic defect is the intestinal zinc-specific transporter gene SLC39A4 on chromosome 8 q24.3 [3]. Initial signs and symptoms usually occur in the first months of life, often after weaning from breast milk, as described in our case, where the rash appeared one month after weaning from breast milk. This has led to assumptions that human milk contains a factor that improves zinc uptake and prevents the disease from being manifested while an

infant is on breast milk [4]. A syndrome resembling acrodermatitis enteropathica was observed in patients with secondary zinc deficiency caused by long-term total parenteral nutrition or by chronic malabsorption syndromes [1].

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Figure 1. [A] Symmetric psoriasiform skin lesion on groin and knees. [B] Similar lesion on buttocks

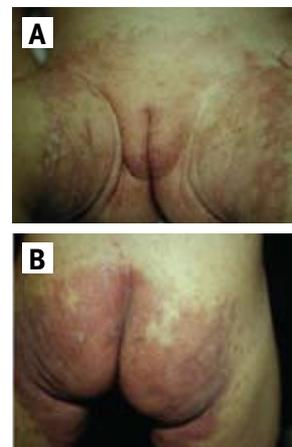


Figure 2. [A & B] Resolution of skin lesions after 2 months of treatment with oral zinc sulfate