

Acrodermatitis enteropathica in a 3-month-old boy

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A 3-month-old boy who had been exclusively breastfed presented with a 2-week history of diarrhea and diaper rash that had not resolved with barrier cream and frequent changes. The infant was born to a 23-year-old primigravida mother at term on the third centile, which he was growing along. His parents were nonconsanguineous, and there was no known family history of genetic conditions. On physical examination, the infant was irritable and had symmetric, sharply demarcated, erythematous, erosive plaques with scaling and crusting on his toes and genital area (Figure 1A), as well as his cheeks, auricles and neck (Figure 1B). We diagnosed acquired acrodermatitis enteropathica. The diagnosis was supported by a low plasma zinc level (18 [normal 70–120] $\mu\text{g/dL}$), and low alkaline phosphatase (20 [normal 96–360] U/L). We started zinc supplementation at a dose of 0.5 mg/kg/d. The infant's symptoms and skin lesions resolved and zinc levels became normal even after zinc supplementation was stopped.

Acrodermatitis enteropathica can be caused by inherited or acquired zinc deficiency.¹ When inherited, it is a result of homozygous mutations in the *SLC39A4* gene, which lead to impaired zinc transmembrane transport and uptake.² It occurs in 1 in 500 000 live births, with no predilection for sex or race.³ Typically, bottle-fed infants manifest the symptoms at age 4 to 10 weeks, whereas breastfed infants manifest the symptoms much later because of increased bioavailability of zinc in breast milk, which also contains a zinc transporter protein that facilitates zinc absorption.¹ Acquired zinc deficiency most commonly arises from zinc-deficient diets or from breast milk deficient in zinc-binding ligand, and affects 2% of children in the United States and 20% in low- and middle-income countries.¹

Physicians should consider measuring serum zinc in infants with irritability, chronic diarrhea, alopecia and typical rash in the periorificial, perineal and acral distribution. Because genetic testing

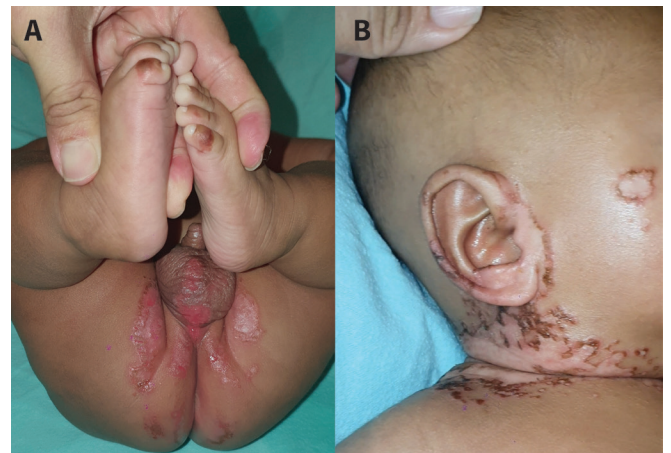


Figure 1: (A) Multiple well-demarcated erythematous erosive plaques in a 3-month-old boy, involving the toes, genital and perineal areas. (B) Well-defined, crusted erosions on the neck and right auricle. Patchy alopecia of the scalp is also visible.

was not available, we made a presumptive diagnosis of acquired acrodermatitis enteropathica based on our patient's marked response to zinc supplementation. In acquired zinc deficiency, improved maternal diet and maturation of the infant's gut means zinc levels often remain normal after supplementation is stopped.

References

1. Leung AKC, Barankin B. Case in point: acrodermatitis enteropathica: an infant with skin lesions and diarrhea from zinc deficiency. *Consultant for Pediatricians* 2016;15:406-8. Available: www.consultant360.com/articles/case-point-acrodermatitis-enteropathica (accessed 2021 Jan. 15).
2. Del Ciampo IRL, Sawamura R, Del Ciampo LA, et al. Acrodermatitis enteropathica: clinical manifestations and pediatric diagnosis [article in Portuguese]. *Rev Paul Pediatr* 2018;36:238-41.
3. Nistor N, Ciontu L, Frasinariu O-E, et al. Acrodermatitis enteropathica: a case report. *Medicine (Baltimore)* 2016;95:e3553.

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