




Acrodermatitis enteropathica with ocular manifestations

Deepsekhar Das , Aishwarya Rathod, Sujeeth Modaboyina , Sahil Agrawal 

Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India

Correspondence to

Dr Sahil Agrawal;
agrawalsahil03.acad@gmail.com

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DESCRIPTION

Acrodermatitis enteropathica is a rare autosomal recessive metabolic disease with occasional ocular manifestations. It was first described by Brandt in 1936 and named so by Danbolt.¹ It occurs worldwide with an estimated incidence of 1 per 500 000 children with no apparent predilection for race or sex.²

A 12-year-old boy presented with reports of chronic diarrhoea, skin lesions, photophobia and purulent discharge from both eyes for a week. Family history was insignificant. On examination, he had acral psoriasis-like dermatitis (figure 1A,B) with cicatricial alopecia (figure 1C). On ophthalmological examination, bilateral cicatricial lagophthalmos with ciliary and eyebrow madarosis was found (figure 1D). The skin over the eyelids was shiny, scaly and hyperpigmented. Ocular surface had conjunctival xerosis and moderate dry eye disease. He had reduced blood levels of zinc (42 µg/dL), and genetic analysis revealed a mutation in the *SLC39A4* gene. He was diagnosed with a case of congenital acrodermatitis enteropathica.

It is a disorder of zinc metabolism with congenital and acquired forms characterised by dermatological manifestations (characteristically vesiculobullous when active), mucosal inflammation (commonly conjunctivitis) and steatorrhea.³ Conjunctivitis, blepharitis and photophobia are the most common affections seen in these cases. The upper part of the cornea is also known to have a band-shaped opacity concentric to the limbus with radiating lines from it.⁴ The disorder is caused by a mutation in the *SLC39A4* gene on chromosome 8q24.3 encoding for a transmembrane protein, which uptakes zinc Zip4 (acts as a transporter).⁵ The metabolism of protein, carbohydrate and vitamins require zinc as an essential trace element, as it is necessary for the proper functioning of all cells.⁶ Zinc replacement therapy should be started at 3 mg/kg/day of elemental zinc (there is 50 mg of elemental zinc per 220 mg zinc sulfate) in acrodermatitis enteropathica. Clinical improvement is seen within days to weeks, quite rapidly, typically even before a significant change in serum zinc levels.⁷ Zinc most characteristically causes gastric irritation with nausea, vomiting and gastric haemorrhage. Multisystem organ failure, endangering life, may be caused by large accidental doses of zinc.⁸

Our patient was started on oral zinc gluconate 3 mg/kg and vitamin A supplementation with monitoring of zinc plasma levels every 3 months and preservative-free lubricants for ocular relief. The



Figure 1 Clinical photograph of the patient showing erythema, scaling and crusting over hand and feet along with nail dystrophy (A,B), cicatricial alopecia over scalp (C) and bilateral cicatricial lagophthalmos with ciliary and eyebrow madarosis (D).

patient improved drastically within a week and was kept on lifelong zinc therapy.

Diagnosis of acrodermatitis enteropathica is based on clinical symptoms (triad of periorificial dermatitis, alopecia and diarrhoea) and low plasma levels of zinc, with a rapid clinical response to supplementation as a confirmation of the disease. Early diagnosis is essential for preventing complications both ocular and systemic.

Patient's perspective

I pray no one else gets such a disease. With treatment I am much better and relieved of my eye problems.

Learning points

- ▶ Although acrodermatitis enteropathica is treatable, early recognition and prompt start of treatment are extremely important.
- ▶ Supplementation of zinc and serum level monitoring every 3 months with lubricants for ocular relief is the key to ophthalmological treatment.



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ORCID iDs

Deepsekhar Das <http://orcid.org/0000-0002-4446-0274>

Sujeeth Modaboyina <http://orcid.org/0000-0002-1306-5722>

Sahil Agrawal <http://orcid.org/0000-0001-6667-249X>

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