CASE REPORT

Acrodermatitis enteropathica-like skin lesions in a neonate

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SUMMARY

A male neonate was born to a sixth-gravida mother with a history of four early-neonatal deaths. On day 21 of life, the patient was admitted for poor feeding, vomiting and encephalopathy. Final diagnosis of propionic acidaemia (propionylcarnitine, 17.67 $\mu\text{mol/L}$) was made. He was managed by peritoneal dialysis followed by protein-free and special lipid diet, sodium benzoate and multivitamins. On day 28 of life, he developed acrodermatitis enteropathica-like skin lesions on perioral and diaper area that did not respond to oral zinc or antimicrobials. A possibility of acrodermatitis acidaemia was kept and supplementation with essential amino acids started, following which the skin lesions regressed.

BACKGROUND

Branched-chain organic acid disorders are autosomal recessive disorders that appear during the first weeks of life, presenting with metabolic acidosis and hyperammonemia. Treatment aims to reduce the accumulation of toxic intermediates by restriction of natural protein. The amino acid-modified formulas required can lead to iatrogenic amino acid deficiencies and cause a skin rash resembling acrodermatitis enteropathica that is known as acrodermatitis acidaemia. Classically, acrodermatitis enteropathica is caused by zinc deficiency, but it is a rare condition nowadays.¹ As acrodermatitis acidaemia is rarer than acrodermatitis enteropathica and commonly children are treated with zinc supplements before the condition worsens, clinicians should be aware of this condition while treating any child with an underlying metabolic disorder including the branched-chain organic acid disorders.

CASE PRESENTATION

A male neonate was born to a fifth-gravida mother with history of four undiagnosed early-neonatal deaths. He was asymptomatic during early-neonatal period and discharged on breast feeds. On day 10 of life, he got readmitted with history of poor feeding, vomiting and lethargy. On examination, he was sick looking, but with stable vital parameters and features of encephalopathy. Investigations ruled out neonatal sepsis. Initial metabolic work-up revealed hyperammonemia (375 $\mu\text{mol/L}$) along with metabolic acidosis. He was started on intravenous fluid containing 10% dextrose, intralipids/multivitamins and sodium benzoate. Peritoneal dialysis led to improvement in clinical and biochemical parameters. Protein supplementation was gradually started. Metabolic workup revealed propionic

acidaemia with elevated propionylcarnitine level of 19.67 $\mu\text{mol/L}$ (normal <7 $\mu\text{mol/L}$). Sodium benzoate was stopped, and the infant was started with carnitine supplementation (200 mg/kg twice daily). Protein supplementation was modified to include a reduced isoleucine, methionine, threonine, valine and odd chain fatty acid content. The infant was discharged with proper dietary advice. On day 40 of life, he presented with desquamating erythematous macules and papules with scaling on perioral, acral and diaper areas, which was not responding to oral zinc or antimicrobials. There was a history of fever along with poor oral intake 7 days prior to the development of skin lesions. The infant was admitted in another hospital, and treated only with intravenous fluids/antibiotics, without any protein/calorie supplementation. The parents were also non-compliant to the dietary advice, while the infant was at home. Instead they were giving some homeopathic medications. The lesions gradually worsened with extensive skin peeling (figures 1 and 2).

INVESTIGATIONS

The infant was worked up to find out the cause. Fungi and bacteria cultures were negative. The serum zinc level was normal. There was deficiency in isoleucine (7 nmol/mL; normal=25–110), valine (54 nmol/mL; normal=110–280), methionine (3 nmol/mL; normal=10–60) and threonine (48 nmol/mL; normal=90–260). A diagnosis of 'Acrodermatitis acidaemia' was made based on compatible skin lesions along with documented deficiency of specific amino acids.

Figure 1 Sharply demarcated desquamating erythematous patches and plaques of dry, scaly skin on perioral area.

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Figure 2 Similar lesions in diaper area.

DIFFERENTIAL DIAGNOSIS

- ▶ Psoriasis
- ▶ Atopic dermatitis
- ▶ Acrodermatitis enteropathica
- ▶ Arginine deficiency

TREATMENT

Diet was established with an increased caloric ingestion and higher amounts of natural proteins in order to increase the essential amino acids.

OUTCOME AND FOLLOW-UP

The skin lesions improved within 48 h, and got completely resolved in a week. Levels of the amino acids got normalised in the repeat serum sample. The infant is currently in follow-up and doing well.

DISCUSSION

Acrodermatitis acidemica refers to cutaneous lesions described in patients with branched-chain organic acid disorders, and sometimes in glutaric aciduria type I.^{1 2} It resembles the skin lesions described in acrodermatitis enteropathica.^{2 3} It may appear in the illness context or as a result of deficiency of

essential amino acids during dietetic treatment.^{3 4} Conditions like prolonged starvation, insufficient intake of calorie/protein, protein-free instead of specific amino acid restricted diet can lead to development of these cutaneous lesions.⁵ In the present case, dietary restrictions along with the infant's illness might have precipitated the skin lesions. Sometimes associated diarrhoea and anaemia might be present. These symptoms also respond to supplementation with essential amino acids, as the latter are essential for cells with rapid turnover, including keratinocytes, bone marrow erythrocyte progenitors and enterocytes.

Learning points

- ▶ Presence of acrodermatitis enteropathica like lesions in children with aminoacidopathies during dietary restriction should raise suspicion of acrodermatitis acidemica secondary to amino acid deficiency rather than a zinc deficiency.
- ▶ Establishment of the diagnosis requires documented normal serum zinc level.
- ▶ Treatment involves an increased caloric ingestion and higher amounts of natural proteins, in order to increase the essential amino acids.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Niiyama S, Koelker S, Degen I, *et al.* Acrodermatitis acidemica secondary to malnutrition in glutaric aciduria type I. *Eur J Dermatol* 2001;11:244–6.
- 2 De Raevé L, De Meirleir L, Ramet J, *et al.* Acrodermatitis enteropathica-like cutaneous lesions in organic aciduria. *J Pediatr* 1994;124:416–20.
- 3 Domínguez-Cruz JJ, Bueno-Delgado M, Pereyra J, *et al.* Acrodermatitis enteropathica-like skin lesions secondary to isoleucine deficiency. *Eur J Dermatol* 2011;21:115–16.
- 4 Domínguez-Cruz J, Bueno-Delgado M, Bernabeu-Wittel J, *et al.* Acrodermatitis acidemica: experience of 5 years in Andalusia. *Pediatr Dermatol* 2010;27:218–19; author reply 219–20.
- 5 Happle R. Acrodermatitis acidemica-ein kutanes Leitsymptom verschiedener Organoazidamien. *Z Hautkr* 2001;76:205.

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