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Acrodermatitis Enteropathica: A Case Report

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Abstract
We present a case of acrodermatitisenteropathica (AE) in a 28 day newborn who came with periorificial and acral dermatitis with diarrhea and had rapid improvement with zinc therapy after 6 days of treatment. We couldnot measure serum zinc level in the patient as it was not available in our centre. Though the treatment is simple and rapid, it is quite distressing and leads to mortality if not treated on time. So, a high index of suspicion is mandatory if there are clinical symptoms that mimics AE so that it can be treated on time.
Keywords*: acrodermatitisenteropathica, zinc deficiency, periorificial and acral dermatitis*

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I. Introduction

Acrodermatitisenteropathica (AE) is a rare genetic autosomal recessive disorder, which is caused due to the mutation in the gene that encodes a membrane protein that binds zinc.[1]The genetic defect has been mapped to 8q24 and the defective gene identified as SLC39A4, which encodes the zinc transporter Zip4.[2]

The triad of AE comprises of dermatitis, alopecia, and intractable diarrhea as its diagnostic hallmark, and is recorded only in 20% of cases. Besides other uncommon manifestations in the form of mood changes, anorexia and neurological disturbances are prominent in infancy, whereas growth retardation, behavioral changes, weight loss, and recurrent infections are prevalent in toddlers and in schoolchildren.[3]

The cutaneous findings are highly characteristic and often present initially as a non specificacrallydistributed, symmetric, eczematous dermatitis, over time, bullae and erosions with a characteristic peripheral crusted border develop. Patients also appear to be predisposed to systemic infections as a result of impaired cell mediated.[4]

II. Case Report

A 28day newborn, female child, born from non-consanguineous parents, via normal vaginal delivery at full term presented with generalized erythematous plaques with eczematization. Birth history was normal and the baby was on exclusive breast feeding and had a weight of 3.5kg with the normal anthropometric measurements. Her skin was apparently normal till 10 days of life.

At the time of presentation to us skin examination revealed erythema with peeling of skin and brownish crusted plaques present over gluteal region, groin area extending upto bilateral thighs, legs, upper back and lower abdomen with relative sparing over anterolateral region of bilateral legs as shown in figure1 and figure 2.Similar lesions present over neck extending upto anterior chest and upper back and multiple satellite lesions over midback. Perioral fissuring and crusting was present with multiple erythematous papules over bilateral cheeks as shown in figure 3. Palms and soles showed exfoliation. Nails were normal with mild paronychial inflammation. There was no skin tenderness and nikolsky's sign was negative. There was no involvement of mucosa. Baby also had loose greenish stools 6-7 times per day for 10 days. There was no history of fever.

Fig1. Peeling of skin with crusting extending from abdomen upto bilateral lower legs

Figure 2. peeling of skin and crusting present over back, gluteal region extending upto bilateral legs

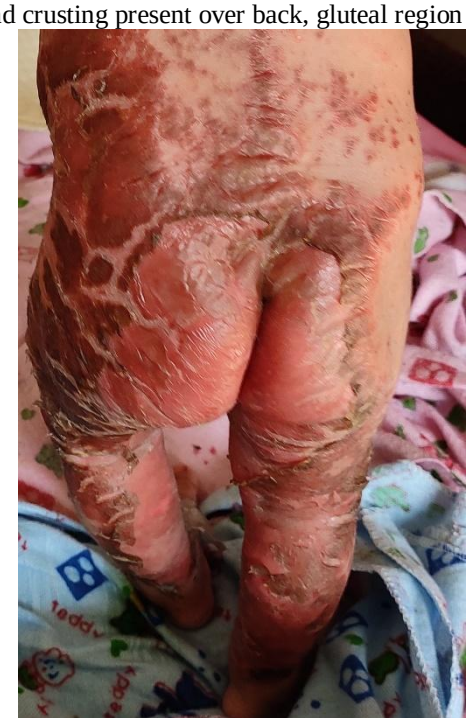
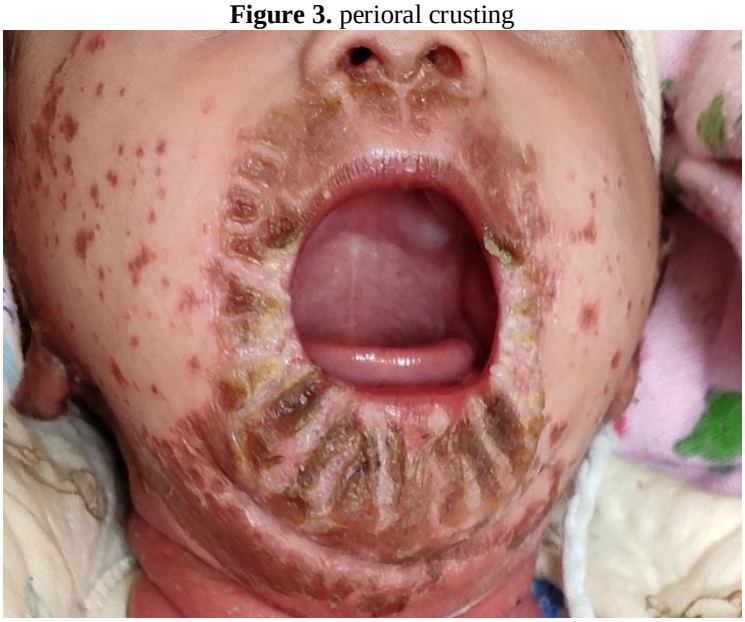


Figure 3. perioral crusting



Family history of similar illness present. Mother of the child had similar lesion when she was a child. Similarly, aunt of the baby and two first cousins also were diagnosed with acrodermatitisenteropathica with improvement of condition with zinc supplementation.

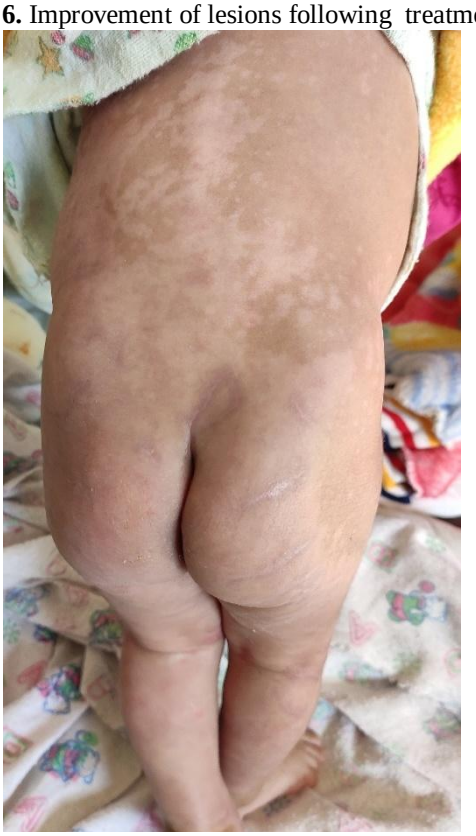
Child was irritable and there was difficulty in feeding. However, there was no history of hair fall.Child was taken to a local hospital where betamethasone was given with the diagnosis of eczema and was applied over skin lesions for 10days which further aggravated the lesions. Child was given BCG vaccine at birth and other vaccinaton were planned according to EPI schedule of Nepal.

Patient was diagnosed with acrodermatitisenteropathica according to the clinical features, skin examination and family history. There was no facility of testing serum zinc level in our centre so couldnot be done. Patient was treated with zinc tablet at the dose of 3mg/kg and a emollient was given to be applied over skin lesions. Rapid improvement of lesions was present after 2 days and on 6th day follow up the lesions were completely subsided over whole body as shown in figure 4 and 5. The frequency of stool also decreased on 3rd day of treatment. Child was playful, active and started feeding well.

This case is reported because though serum zinc level couldnot be measured, clinical evaluation and skin findings can definitely be helpful in diagnosing this rare condition and there is rapid improvement of the condition with oral zinc supplementation. Therefore, this case emphasizes the need for early diagnosis and prompt treatment required to reverse the condition, reduce mortality and prevent the long term consequences of zinc deficiency.

Fig 5. Improvement of lesions after treatment

Fig 6. Improvement of lesions following treatment



III. Discussion

Acrodermatitisenteropathica can be congenital or acquired and the prevalence of congenital AE is approximately 1 in 500,000 children. In congenital AE, there is defect in active zinc transport in the duodenal mucosa.[5]

AE is usually reported in premature babies due to insufficient storage of zinc as well as high zinc requirements, and at the time of weaning. However, it is rare in breastfed infants as in our case. The presence of zinc binding ligand in breast milk increases bioavailability of zinc thus protecting breast fed infants.[6]

The cutaneous lesions are psoriasiform, erythematous, scaly and crusted plaques. As the disease progresses, these lesions may become vesicobullous, pustular and erosive. Similarly other features that may be present include stomatitis, apathy, irritability, growth retardation, failure to thrive and delayed wound healing.[7]In our patient, characteristic perioral crusting and acrally distributed plaques were present along with diarrhea.

Laboratory diagnosis is hazardous, zinc levels in serum, urine, or hair are used (although they are not specific, neither sensitive), zinc absorption tests are cumbersome, genetic testing is definite (defect in 8q24, gene SLC39A4) though not routinely available. Most clinicians therefore depend on immediate results of a therapeutic zinc regimen (3–30 μmol/kg body weight).[8] Like in our case, serum zinc level couldnot be done as it was not available in our centre and patient party refused to do it by sending sample to other labs due to poor financial status.

Histopathological examination of the skin is neither specific nor diagnostic. If the patient presents with acrodermatitisenteropathica-like skin lesions in the perioral and diaper area, not doesnot respond to oral zinc, other conditions like biotin deficiency, essential fatty acid deficiency, phenylketonuria, organic acidemias, and metabolic disorders of the urea cycle like glutaricaciduria type I, ornithine transcarbamylase deficiency, and citrullinemia should be considered.[6]

References

[1]. Nistor N, Ciontu L, Frasinariu OE, Lupu VV, Ignat A, Streanga V. Acrodermatitisenteropathica: a case report. Medicine. 2016 May;95(20). https://doi.org/10.1097/MD.0000000000003553

[2]. Maverakis E, Fung MA, Lynch PJ, Draznin M, Michael DJ, Ruben B, Fazel N. Acrodermatitisenteropathica and an overview of zinc metabolism. Journal of the American Academy of Dermatology. 2007 Jan 1;56(1):116-24. https://doi.org/10.1016/j.jaad.2006.08.015

[3]. Sehgal VN, Jain S. Acrodermatitisenteropathica. Clinics in dermatology. 2000 Nov 1;18(6):745-8. https://doi.org/10.1016/S0738-081X(00)00150-4

[4]. Shakya NB, Rajbhandari SL, Jha SM. Acrodermatitisenteropathica: a case report. Medical Journal of Shree Birendra Hospital. 2011;10(2):32-4. https://doi.org/10.3126/mjsbh.v10i2.6461
[5]. Karadag AS, Bilgili SG, Calka O. Acrodermatitisenteropathica in three siblings. Indian Journal of Dermatology, Venereology, and Leprology. 2013 Mar 1;79(2):268. https://doi.org/10.4103/0378-6323.107667
[6]. Pinto M, Bhat MR, Dandekeri S, Kambil SM. AcrodermatitisEnteropathica in an infant with normal zinc levels. Indian Journal of Dermatology, Venereology, and Leprology. 2015 Jan 1;81(1):70. https://doi.org/10.4103/0378-6323.148583
[7]. Agarwal S, Gopal K. Acrodermatitisenteropathica in a breast-fed infant. Indian Journal of Dermatology, Venereology, and Leprology. 2007 May 1;73(3):209. https://doi.org/10.4103/0378-6323.32757
[8]. Nistor N, Ciontu L, Frasinariu OE, Lupu VV, Ignat A, Streanga V. Acrodermatitisenteropathica: a case report. Medicine. 2016 May;95(20). https://doi.org/10.1097/MD.0000000000003553

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