

**PRIMARY ACRODERMATITIS ENTEROPATHICA**

***¹DR RUGMINI KAMALAMMAL, ²DR. S. SANTHA KUMAR, ³DR. GUNASEKARAN ANANDAN,
⁴DR. ABDUL AZIZ.A AND ⁵DR. SANGEETHA HARIPRASATH**

*¹ Associate Professor Department of paediatrics Sree Balaji Medical College and Hospital,
Bharath University Chrompet Chennai – 44*

*^{2,3,4,5} Junior Resident Department of paediatrics Sree Balaji Medical College and Hospital
Bharath University Chrompet Chennai – 44*

ABSTRACT

Acrodermatitis Enteropathica is characterized by erythematous patches, plaques, pustular and bullous lesions with acral and periorificial distribution, diarrhoea and abnormally low Zinc levels. We are presenting a baby with the characteristic rash, failure to thrive and diarrhea with reduced levels of Zinc. The baby was treated initially for lactose intolerance but lesions did not subside. There was complete resolution with therapeutic dose of Zinc supplementation and emollients. The mother's serum zinc levels were extremely low suggesting a diagnosis of primary Acrodermatitis Enteropathica. The case is being presented for highlighting the rare primary form of Zinc deficiency and its dermatological features.

KEY WORDS : Acrodermatitis enteropathica, Zinc deficiency

**DR RUGMINI KAMALAMMAL**

D97, Sunnyside Apartments, Konnur High Road, Ayanavaram, Chennai – 23. Associate Professor Department of paediatrics Sree Balaji Medical College and Hospital Bharath University Chrompet Chennai – 44

*Corresponding author

INTRODUCTION

Zinc deficiency is characterized by dermatitis with an acral and periorificial distribution, of pustules and bulla, erythematous plaques, dry scaly lesions or eczema like lesions. associated with alopecia of the scalp and eyebrows, thin and friable hair, paronychia, angular stomatitis, glossitis and diarrhoea^(1,2) Acute phase lesions are erythematous papules, which may turn into pustules and may show exudation. Subacute lesions are usually dry erythematous excoriated papules.⁽³⁾ Acrodermatitis Enteropathica is a disorder due to zinc deficiency, it can either be primary autosomal recessive disorder or secondary Acrodermatitis Enteropathica due to acquired zinc deficiency following lactose intolerance, cow's milk protein allergy, cystic fibrosis etc. The incidence is 1/ 50,000 children Primary Acrodermatitis Enteropathica is due to mutations in the gene [SLC39A4] located in chromosome 8q24 which encodes zinc transporter belonging to ZIP family in humans.^(2,4) About 30 mutations are reported.⁽⁴⁾ Treatment of this condition is by Zinc supplementation of 1-3 mg /day of elemental zinc and it is to be continued life long, in case of primary deficiency.

CASE REPORT

A 3 months old female child presented to us with failure to thrive with erythematous skin lesions. The baby was a term baby born to non consanguineous parents with birth weight of 3.3kg. Mother gave a history of persistent diarrhea since 1 month of age associated with bullous skin lesions, and it was diagnosed by a

local physician as lactose intolerance, and was advised to stop breast feeding. The baby was started on soya based formula feeds, and diarrhea subsided within a few days but the skin lesions were still present. After 2 months of soya based formula feeds it was noticed that baby was not gaining weight and developed diarrhea, erythematous plaques with scaling over the peri anal region, inguinal region, thigh, forearm and neck region. There was no associated itching. Some of the lesions evolved into blisters. On applying emollients, these lesions turned into hypopigmented, well demarcated patches. The weight of the baby was 3.6kg which was less than the 3rd percentile for the age. Systemic examination was normal except for a ulceration over the BCG scar site which also turned into a scab after one month. Complete blood count done was normal, serum zinc level was 58 microgram/dl [normal being 70 to 150 microgram/dl]. Serum IgG was within normal limits. Mother's serum zinc level was 34 microgram/dl [method ECLIA], which is significantly lower than the normal limits. Both baby and mother were found to be HIV negative. The 3 elder siblings of the baby were normal. There was no family history of similar illness. The baby was treated with oral zinc acetate 40mg/day (therapeutic dose), emollients, the formula feeds were given in a correct dilution. Baby gained weight over a period of 2 months, the lesions resolved. Skin biopsy was not done as the parents were reluctant to do an invasive procedure for the baby.

BABY SHOWING SKIN LESIONS

Before Treatment



After Treatment



DISCUSSION

The congenital form of Acrodermatitis enteropathica usually presents on weaning or on stopping breast milk. ⁽⁴⁾Increased bioavailability of Zinc in breast milk gives protection for breast fed babies ⁽⁵⁾. Phenotypic variability is observed in some patients ⁽⁴⁾. Congenital form presence with gross reduction in serum zinc level, but the acquired form usually shows only a marginal deficiency ⁽³⁾. Congenital form is diagnosed when the triad of clinical symptoms, namely, acral dermatitis, alopecia, diarrhea is present with low serum Zinc level. In this form recurrence occurs on stopping Zinc supplementation. Our case fits into primary Acrodermatitis Enteropathica, as the baby had diarrhea, dermatitis and both mother and baby had zinc deficiency. ⁽⁶⁾ The diarrhea in this baby was attributed first to lactose intolerance. Even though there was an

initial response, the recurrence of diarrhea and persisting skin lesions while on soy based formula, rules out lactose intolerance as the primary cause. The relapse and its response to therapeutic doses of Zinc clearly suggests Zinc deficiency as the primary cause. The failure to thrive may be due to diluted soy based formula feeds, and also partially due to persistent zinc deficiency. The ulceration in the BCG scar site is attributed to reduced immunity due to malnutrition. Mother had stopped lactation and food challenge with lactose was not given as it is not advisable below 6 months of age. The mother's serum Zinc levels were extremely low but she was free of any clinical symptoms, and it can be attributed to phenotypic variability ⁽⁴⁾. Probably, the breast milk bioavailability of Zinc was less in this case leading to onset of symptoms during breast feeding period. We

could not do a genetic analysis due to logistic reasons. A genetic analysis of the baby and both parents would have confirmed whether the inheritance is autosomal recessive. Lack of symptoms in the mother might be explained by nonsense mediated decay targeting the mRNA produced⁽⁴⁾. Authors feel that there is a possibility autosomal dominant variant in this condition as here the mother's zinc level was extremely low. This hypothesis is to be studied further. Atopic dermatitis and food allergy are possible explanations for this lesion as these conditions mimic each other. But the Total

serum IgE which is a marker of atopy is normal in this case. In case of atopic dermatitis and specifically in food allergy the IgE is very high⁽⁷⁾

CONCLUSION

Prompt recognition of the disorder early when the child presents with typical skin lesion is essential as therapeutic dose of Zinc gives a good prognosis.

REFERANCES

1. Mortimer PS, Gough P, Newbold PC, Dawber RP, Ryan TJ. Acrodermatitis enteropathica. *J R Soc Med*, 77: 67–68. (1984)
2. Park CH, Lee MJ, Kim HJ, Lee G, Park JW, Cinn YW. Congenital zinc deficiency from mutations of the SLC39A4 gene as the genetic background of acrodermatitis enteropathica. *J Korean Med Sci* 25 (12):1818-20. (2010)
3. da Matta Ain AC, dos S Valente E, Mallozi MC, Sarni RO, Furquim M, Solé D. Acrodermatitis enteropathica-like simulating severe atopic dermatitis: a case report. *Allergol Immunopathol (Madr)*, 36(3):176-9, (2008)
4. Kilic M, Taskesen M, Coskun T, Gürakan F, Tokatli A, Sivri HS, Dursun A, Schmitt S, Küry S. A Zinc Sulphate-Resistant crodermatitis Enteropathica Patient with a Novel Mutation in SLC39A4 Gene. *JIMD Rep*:25-8. (2012)
5. Agarwal S, Gopal K. Acrodermatitis enteropathica in a breast-fed infant. *Indian J Dermatol Venereol Leprol*. 2007 May-Jun; 73(3):209
6. Azevedo PM, Gavazzoni-Dias MF, Avelleira JC, Lerer C, de Sousa AS, Azulay DR. Acrodermatitis enteropathica in a full-term breast-fed infant: case report and literature review. *Int J Dermatol*, 47(10):1056-7, (2008)
7. Diana P, Martin, Nutthapong Tangsinmankong, John W. Sleasman, Noorbibi K. Day-Good, Danita R. Wongchantara. Acrodermatitis enteropathica-like eruption and food allergy. *Annals of Allergy, Asthma & Immunology*, Vol 94 (3) , 398-401. (2005).