Bullous presentation of acrodermatitis enteropathica in three female siblings: A case series

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I. Abstract Body

Zinc is an essential trace element that plays a role in growth and development. Its deficiency results to an autosomal recessive disorder called acrodermatitis enteropathica caused by a mutation of the *SLC39A4* gene encoding the zinc transporter. Clinical and histopathologic findings in literature are diverse often leading to a delay in the diagnosis and treatment. The disease is known for its phenotypic triad of acral and periorificial dermatitis, alopecia, and diarrhea which can be resolved by zinc supplementation.

This is a case series of three female siblings who came in for consult due to periorificial and acral eroded plaques which initially presented as bullous lesions. Patients also manifested diffuse alopecia, nail dystrophy, failure to thrive, irritable mood, and stunted growth. History of consanguinity was present in the parents. An initial assessment of epidermolysis bullosa was made but histopathologic findings were non – specific for an autoimmune blistering disease. Clinically, the patients were diagnosed as acrodermatitis enteropathica and this was confirmed with markedly decreased levels of serum zinc. They were successfully treated with oral zinc sulfate at a dose of 5mg/kg/day for the first two weeks then maintained on a dose of 2mg/kg/day.

II. Image:



III. Reference:

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