Acrodermatitis Enteropathica: Case Report

Asma Al Naamani[1] and Tuqa Al Lawati[2]*

[1]Dematology Department, Al Nahdha Hospital, Muscat, Sultanate of Oman

[2]Medical officer, Ministry of Health, Muscat, Sultanate of Oman

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**Corresponding author tuqa42@gmail.com*

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Abstract

Acrodermatitis enteropathica is a rare autosomal recessive disease which is caused by genetic mutation leading to zinc deficiency. Clinical manifestation includes skin lesions, diarrhea and alopecia. We report a 2-month-old girl, admitted with erythematous scaly lesions in the neck and vesiculopustular lesions in the perioral region, associated with alopecia and diarrhea. Clinical diagnosis of the disease was made from her first presentation. Zinc level was sent and she was started on zinc therapy as recommended and her lesions resolved completely after 1 month of treatment.

Introduction

Acrodermatitis Enteropathica (AE) is a rare autosomal recessive disorder which affects 1 in 500,000 children without prediction of race and gender. It is caused by zinc malabsorption in the intestines. Symptoms defer according to the age, but the main clinical manifestations include diarrhea, alopecia cutaneous lesions and recurrent infections [(1)]. Zinc level in the serum and urine might help in the diagnosis but the definite diagnostic test is genetic testing of the gene mutation. Treatment include mainly of zinc therapy which depends on the level and the body weight [(2)].

Case report

We present our case of 2-month-old girl who was born at 33 weeks of gestation by cesarean section due to maternal severe pre-eclampsia with a weight of 1.98Kg. She was admitted in the neonatal intensive care unit for three weeks and since then she was healthy on breastfeeding. She was

brought by her mother to the accident and emergency department with 1-month history of skin lesions which involved the perioral region, hands and skin folds including napkin area. This was associated with cracked lips, hair loss and diarrhea of more than 7 times per day. The mother noticed that her daughter was excessively crying, lethargic and was febrile. Patient was admitted previously in another hospital with superinfected skin lesions which was treated with intravenous antibiotics for four days.

At the initial assessment of the patient, she looked lethargic and dehydrated. Skin examination revealed; erythematous scaly patches on the neck and nappy area as well as vesiculopustular lesions predominantly on the perioral region (Figure 1). Alopecia was noticed as well. We considered the diagnosis of Acrodermatitis Enteropathica (AE). Patient was hospitalized, laboratory tests were sent which showed normal inflammatory markers, anti-tissue trans glutamine for celiac disease, B-lactoglobulin for cow's milk protein intolerance and immunoglobulins but low alkaline phosphatase (45 IU/L). Zinc level was sent to France and we were awaiting the result. While waiting for blood culture, urine cultures and swabs from the lesions, patient was started on intravenous cloxacillin, cefotaxime and zinc therapy was instituted at a dosage of 3 mg /kg daily of elemental zinc. During the admission, patient started improving in terms of the diarrhea which had stopped, skin lesions started to become paler and smaller after 3-4 days. All cultures and swabs resulted as no growth. She completed 14 days of IV antibiotics and discharge home to continue daily zinc treatment with a follow up appointment at Pediatric dermatology clinic. Patient did not do the genetic testing as it is only available in one institute and difficult to be accessed for all patients.

At the follow up appointment, Zinc level was resulted as < 1.9umol/l. The patient completely improved, and the skin lesions and alopecia have completely remitted (Figure 2). Dose was adjusted to 0.5 mg/kg/ day according to her weight. She is continuing her zinc treatment and will continue regular check-ups with pediatric gastroenterology and dermatology clinic at the regional hospital.

Figure 1: Erythematous scaly lesions in the neck (A), Perioral erythematous vesiculopustular lesions (B).

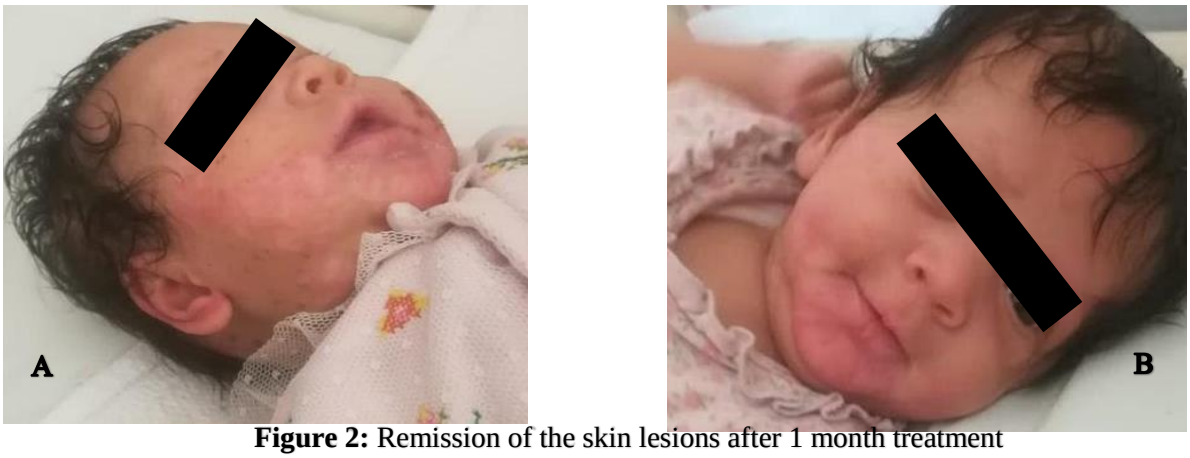

Figure 2: Remission of the skin lesions after 1 month treatment

Discussion

Acrodermatitis enteropathica is an autosomal recessive disorder, which is mainly caused by zinc deficiency. Zinc is an essential element in the body which has a great role in human immune system, growth and development, hormones synthesis and activation as well as gene regulation [(3)]. It is also considered as co-enzyme for several enzymes including alkaline phosphatase, alcohol dehydrogenase, RNA polymerase and numerous digestive enzymes [(4)].

Zinc deficiency (ZD)can be classified into inherited (genetic) and acquired. Patients who are at high risk of developing acquired ZD are those with malabsorption disorders such as celiac and crohn's disease, premature, low birth weight and those who received exclusive parenteral feeding or who are on low zinc breast milk feeding [(5)]. Genetic ZD, also called AE is mainly due to mutation in SLC39A4 gene localized in 8q24.3 which encodes zinc/iron regulated transporter-like protein 4 (ZIP-4)[(1)(2)]. This protein facilitates zinc absorption in the intestine and its metabolism in the body [(2)]. Our patient was born with very low birth weight of 1.98Kg and she was premature as well which put her on high risk of developing zinc deficiency.

The clinical presentation of AE defers according to the age group however the classical triad of the disease involve; alopecia, Periorificial dermatitis and diarrhea, which only accounts for 20% of all patients with AE which our patient presented with. Infants usually present with diarrhea, neurological symptoms and anorexia, while school children and toddlers experience growth retardation, alopecia, weight loss and recurrent secondary infections [(6)]. Severity of skin lesions might defer from an eczematous erythematous rash to vesicular to bullous rash however, early recognition and treatment should be the main goal to prevent further deterioration of the disease

[7]. In our patient, the rash was eczematous in the neck and napkin area while it appeared pustular in the perioral region while study by Kaur S et al' mentioned that patient had only eczematous moist plaques distributed over the perioral and perineal region as well as the buttocks [2].

In our case, patient was diagnosed immediately with acrodermatitis enteropathica because she presented with typical perioral dermatitis, diarrhea and alopecia. However, if patient presented with vague signs and symptoms, other differential diagnosis including atopic dermatitis, contact dermatitis, seborrheic dermatitis, psoriasis and malabsorption disorders should all be excluded [8]. Serum zinc level; normal value (12.2 to 18.2 µmol/L) is the mostly used investigation along with alkaline phosphatase (ALP) level; normal value is (44 to 147 IU/L). In our patient, both serum zinc level and ALP were low while in another study ALP level were normal [2].

AE should be followed up lifelong but the great thing about this disorder is that once it is diagnosed and treatment has been started, symptoms will disappear within few days with survival rate will be 100% [2]. Recommended elemental zinc therapy is 3mg/kg/day as initial treatment followed by 1-2mg/kg/day as a maintenance dose [9]. Follow up is required for adjustment of the dose according to the weight and to check for zinc toxicity as it was done with our patient. Studies have shown that high zinc levels might lead to hypercupremia resulting in alteration of the immune system and further complications [10].

Conclusion

Acrodermatitis enteropathica is a rare condition which needs early recognition and management of the patient. As per our case which presented with typical signs and symptoms, diagnosis was made early and treatment was started according to the recommended regimen. Following up those patients is necessary to avoid zinc toxicity, adjust the dose according to patient weight and to recognize the improvement to maintain the best quality of life.

Acknowledgments

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Limitations:

The only limitation we had in our case study is the access to genetic testing as it available in one institute and difficult to be accessed by all patients all over the country.

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