Chapter 10
Clinical Research Case Descriptions

10.1 Patient One

The patient was a 21-month old Caucasian boy with normal birth weight who had a history of acral lesions and periorificial dermatitis since he was 3 months old. He was breastfed until he was two and a half months old. He has a history of upper respiratory tract infections and otitis media which was treated in a private clinic. His parents were blood-related and his older brother, who had the same symptoms, had died at the age of 14 months. His preliminary skin examination showed plaque like scaly skin, erythematous, dry skin, and skin lesions on areas around the eyes, mouth, and diaper area. Additionally, there were reports of hair loss (alopecia), soft tissue infections around the nails (paronychia), and cracks and infection in the inguinal region. Physical examinations stated that the infant irritable and photophobic. His measured height and weight were 68 cm and 7.8 kg, respectively.

The infant patient was admitted to another hospital because of his chronic intermittent diarrhea, where his skin lesions were cured by the treatment of topical steroid and antifungal therapy. However, the disorder was not cured and the symptoms had not improved.

The patient was hospitalized at Razi Hospital when he was brought in for the evaluation of his skin lesions, which were caused by Staphylococcus aurous and septicemia caused by

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Klebsiella. He was treated with intravenous antibiotic therapy and after a few days his general condition improved, except for his skin lesions. The blood and urine tests were normal but his plasma zinc level was measured to be at 12 μg/dL (normal levels are 50–150 μg/dL). Moreover, microscopic examinations showed Candida albicans on the inguinal area, which was treated in 12 days by the administration of Baflokonazol 6 mg/kg/day. The patient was diagnosed with Acrodermatitis Enteropathica because of his clinical and laboratory test results. He was prescribed 40 mg of oral zinc sulfate per day which resulted in significant improvement of the symptoms within a few days, including the skin lesions. After a month of treatment, all lesions healed, hair began to grow back, and there were no reports of diarrhea. A follow up with the patient was recommended.

10.2 Patient Two

The patient was a 25-year-old woman. When she was 2 months old her mother stopped breastfeeding her and skin eruptions covered her entire body. The skin eruptions, such as vesiculobullous and erythematous scaly lesions, started around her mouth and spread throughout her face and legs. She also had diarrhea. Since she had symptoms suggestive of AE, there were further tests done in order to confirm the diagnosis. The diagnosis was confirmed by measurement of the plasma zinc levels which were at 0.2 mg/L (normal levels are between 0.7 and 1.6 mg/L). Her treatment began with administration of four tablets of 40 mg zinc sulfate per day. After 4 days, there was incredible improvement of her skin rashes and diarrhea, but she had developed behavior disorders. During these 25 years she continued her zinc supplementation treatment.

When she was 15 years of age, her treatment was reduced from 220 mg three times daily to twice daily. However, after 2 weeks, skin lesions began to appear around the mouth. When she was 16 she developed severe vomiting; and as a result, her treatment was increased to 220 mg of spansule-zinc

supplements three times a day again. Afterwards, her plasma zinc was decreased from 1.1 to 0.3 mg/L and she suffered from rashes, similar to eczema and mild seborrheic dermatitis, on the sides of her nose and rashes similar to acne on her body. These rashes began to disappear 3 weeks after the increasing of spansule-zinc supplements to three times a day: plasma zinc increased to 1 mg/L.

Plasma zinc levels were measured at regular intervals of 6 months. When the patient reached the age of 20, her plasma zinc level decreased to the lowest normal level: 0.7 mg/L. The spnasule-zinc treatment was increased to 10 times a day. Fortunately, the patient's body was able to tolerate 10 pills per day and her plasma zinc level was maintained at a normal level of 1.4 mg/L.

Since this medication was not supported by the NHS, the patient had to pay the entire cost of her treatment for a long time. She used Oral Contraceptive Pills (Brevinor) without any effects on AE: OCP has been known as one of the causes of zinc deficiency. In addition the minor increase in zinc demand by the patient's body is because of fetal development. As a result, when the patient was pregnant, the level of plasma zinc was kept constant, since this deficiency disorder may be teratogenic. It is important for plasma zinc levels to be maintained at a normal concentration, because zinc deficiency may result in behavior and mood disorders, such as Cerebellar disorders, Parkinsonism, and cortical atrophy.

10.3 Patient Three

The patient was a 10-month-old female baby when she was administer to Razi Hospital, who presented skin eruptions, including sharply demarcated patches of vesiculobullous dermatitis on the cheeks, fingers, and in the diaper region when she was 2 months old. Her initial skin manifestations appeared 2 weeks after her weaning. She also showed symptoms of recurrent diarrhea, partial alopecia, and growth retardation. She was also observed to be irritable and had photophobia.

It is interesting to note that the patient's parents were not blood-relatives. The patient was born by a full term normal delivery; however since she had symptoms suggestive of AE, there were further tests done in order to confirm the diagnosis. Liver and renal function test results were normal. The diagnosis was confirmed by measurement of the serum zinc levels which were at 35 μg/dL (normal levels are between 70 and 150 μg/dL). Her serum alkaline phosphatase levels were also low at 57 IUL (normal levels are between 115 and 360 IUL).

The patient began substitution therapy with zinc sulfate 5 mg/kg body weight (50 mg daily) which led to the cessation of diarrhea after 2 weeks. The patient's plasma zinc levels were measured at regular intervals of 6 months. During the patient's follow- up after 4 months, the infant exhibited normal development and there was no recurrence of lesions or diarrhea.

10.4 Patient Four

This patient was a 4-month-old female baby when she was administered to Razi Hospital, who presented multiple well-defined erythematous, oozy crusted lesions on hands, feet, and periorificial areas. She developed these lesions 1 month after weaning. She was born by full term normal delivery to non-consanguineous parents. After further clinical examinations it was reported that she had normal anthropometric measurements, but was irritable. Furthermore, there was no report or indication of diarrhea or photophobia.

Since she had symptoms suggestive of AE, there were further tests done in order to confirm the diagnosis. Hematogram, liver, and renal function test results were normal; however, her serum zinc levels were at 48 μg/dL (normal levels are between 70 and 150 μg/dL). Her serum alkaline phosphatase levels were normal at 213 IUL (normal levels are between 115 and 360 IUL).

One week after the administration of zinc sulfate 5 mg/kg body weight (50 mg daily) the cutaneous lesions showed signs of improvement. One month after the administration, her serum zinc level was re-measured and the level was found to

be higher at 102 μg/dL. During discharge, lifelong continuation of oral zinc supplementation was advised.

10.5 Patient Five

A 9-month-old male baby was administered to Razi Hospital after visiting the local clinic with superinfected, sharply demarcated, symmetrical erythematous macules and vesicles. Yellow-coloured crust manifestations were observed on the patient's cheeks as well as on the perioral, perianal, fingers, and diaper region. The baby was born by full term normal delivery and his parents were second degree blood-relatives. The skin lesions had appeared 1 week after weaning and he also showed signs of irritability and photophobia.

The infant's 4-year-old sister was also reported to be affected by similar manifestations and so the initial diagnosis of the clinic physician was a contagious impetigo. However, the suspected contagious impetigo had failed to respond to both topical and systemic antibiotics. Further clinical and laboratory tests were done at Razi hospital. Hematogram, liver, and renal function test results were normal; however, his serum zinc level was at 35 μg/dL (normal levels are between 70 and 150 μg/dL). His serum alkaline phosphatase levels were at 87 IUL (normal levels are between 115 and 360 IUL).

The patient was administered zinc sulfate 5 mg/kg body weight (60 mg daily). After 2 weeks of administration, the skin lesions showed signs of improvement. It was observed that his serum zinc level was found to be higher at 116 μg/dL, when measured 1 month later. Lastly, during discharge, lifelong continuation of oral zinc supplementation was advised.

10.6 Patient Six

A 4-year-old girl, sister of patient five that was described in the previous section presented similar lesions localized on her face, hands, and body folds. She was administered to Razi Hospital

after the suspected impetigo failed to respond to both topical and systemic antibiotics. Dermatological examinations revealed eroded and beige coloured dry patches on the dorsum of the hands, perioral, and inguinal regions. As it was mentioned previously, the girl's parents were second degree blood-relatives.

Further clinical and laboratory tests were done at Razi hospital. Her serum zinc level was measured at 63 μg/dL (normal levels are between 70 and 150 μg/dL) and her serum alkaline phosphatase levels were measured at 280 IUL (normal levels are between 115 and 360 IUL).

The patient was administered zinc sulfate 5 mg/kg body weight, and 2 week later the skin lesions showed signs of improvement.

10.7 Patient Seven

The patient was a 2-month-old male baby, who was administered to Razi Hospital. He presented perioral and acral bullous lesions: the acral sites being the patient's hands and feet. These manifestations were variable sized vesicles and pustules with an erythematous base. The patient was observed to be febrile and irritable. There were no reports or indications of diarrhea or photophobia. He was born by full term normal delivery and was breast fed. The skin lesions appeared two weak after weaning. There was no family history of zinc deficiency.

Since he had symptoms suggestive of AE, there were further tests done in order to confirm the diagnosis. Hematogram, liver, and renal function test results were normal; however, his serum zinc levels were at 43 μg/dL (normal levels are between 70 and 150 μg/dL). This test result was used to confirm the AE diagnosis. His serum alkaline phosphatase levels were normal at 180 IUL (normal levels are between 115 and 360 IUL).

The patient was administered zinc sulfate 5 mg/kg body weight (50 mg daily). After 2 weeks of administration, the skin lesions showed signs of improvement. It was observed that his serum zinc level was improved to 103 μg/dL, when measured 1 month later.

10.8 Zinc Deficiency Similarities to AE in Infants (Eighth Patient)

The patient was a 15-week-old infant with the symptoms of skin rash, loss of appetite, and diarrhea. The skin rash did not respond to topical steroid treatment, antibiotics, antifungal, or protective creams. The patient was born prematurely at 35 weeks with normal skin. His parents were not blood-related and his growth chart was normal. From birth he was exclusively breastfed, and his parents did not report any history of specific disease or disorders. Additionally, the patient's sibling was reported to be healthy and had no history of skin disease. The examinations showed psoriasiform plaques in the groin and peripheral scaling. There were also circular patches observed on both of his cheeks and lips. Dark red psoriasiform plaques were also spotted on the surface of occipital in the posterior hairline, similar to the rashes present around the eyes and mouth. Furthermore, the patient was diagnosed with zinc deficiency since his plasma levels of zinc were at 3 μg/L (normal range is 11–24 μg/L). Oral zinc treatment (22.5 mg/day) resulted in rapid, successful improvements in symptoms: patient's appetite, dermatitis on his chest, other skin eruption, and diarrhea were all showing signs of improvement within 5 days of treatment. This case is suggestive of a transient form of zinc deficiency in and not hereditary AE.