Acrodermatitis enteropathica

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Aim: The aim of the work was the presentation of one case with Acrodermatitis enteropathica. Methods: Acrodermatitis enteropathica is diagnosed based on the pedigree, typical clinical manifestations on the skin, laboratory results, small bowel biopsy, skin biopsy and karyotype. Results: The patient was a two years old male toddler, hospitalized due to skin changes, chronic diarrhoea and total alopecia. Skin changes appeared on akral of the limbs, inguinal and perineal region, joints, perioral area and eyes. These changes appeared in different forms (erythematous, squamous, eczematoid, psoriasisform and crusted). In the eyes were present these changes: blepharitis and conjunctivitis. Also total alopecia was present. Diarrhoea was chronic and specific. Laboratory findings showed the existence of sideropenic anemia, hypoproteinemia with hypoalbuminemia and low plasma zinc concentration (7.5 µmol/L). Hystopathological changes on the small bowel and skin biopsy were not typical for this disease. Following the beginning of treatment with zinc sulphate, all clinical skin manifestations disappeared within two months, but the disease itself was characterized with the periods of exacerbaition and remission. Conclusion: Acrodermatitis Enteropathica is a rare hereditary autosomal recessive disease. Mandatory clinical manifestations are: skin changes, chronic diarrhoea and alopecia. Treatment with zinc is obligatory for the life time. Key words: Acrodermatitis, Diarrhoea, Alopecia.

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1. INTRODUCTION

Acrodermatitis enteropathica is a disease caused by disorder of zinc absorption. It is inherited in autosomal recessive manner, mutant gene SLC39A4 is located on the chromosome band 8q24.3. The global incidence rate is 1:500.000 newborns. Genetic mutation in zinc absorption in the small bowel consists in the specific gene which encodes the transport of zinc SLC39A4. Molecular pathogenetic mechanism consists in the total deficiency of binding factor of zinc, which is produced in pancreas. This factor connects and transports zinc to the epithelial cells of the small bowel. This factor is also secreted in breast milk, therefore the clinical manifestations of this disease are not present among the breastfeeded toddlers (5, 6, 9).

It appears very rarely among the newborns, it does appear among all groups independent of their ethnic origin or sex (2). The disease is characterized by three clinical manifestations: acral dermatitis, diarrhoea and alopecia (3, 12).

2. AIM

The aim of this work was the presentation of one case with Acrodermatitis enteropathica. The patient was treated at the Paediatric Clinic, being at the same time the first case registered in Kosovo.

3. METHODS

For the disease diagnose the following were used: anamnesis, physical examination (described skin changes), plasma zinc concentrations (measured using atomic absorption spectrophotometry), small bowel biopsy, skin biopsy, pedigree and karyotype (was done from peripheral blood lymphocytes).

4. CASE PRESENTATION

Patient (M.R), male, two-year old, weight 10.5 kg (<5percentile), height 82 cm (5-10 percentile), has growth failure. He was a third child from the third pregnancy (Figure 1) and was hospitalized at the Paediatric Clinic of Prishtina during 2008 due to some skin changes and chronic diarrhoea. The skin changes started to appear subsequently with feeding the toddler with the cow’s milk, when the toddler...
Acrodermatitis enteropathica was 10 months old, till the time of his hospitalization the patient was treated as Atopic dermatitis. Skin changes appeared with the symmetric distribution on periorificial areas, acral parts of the limbs, elbows, knees, ankles, inguinal and perigenital area and on the back (Figure 2, 3). Skin changes appeared in different forms: erythematous, vesicular, psoriasiforme and eczematoid and crusted, but not rarely these changes appeared in combination with each other (Figure 2). Manifestations in the eyes were: blepharitis, conjunctivitis, and photophobia, whereas total alopecia appeared on the head. (Figure 4).

Chronic diarrhoea appeared 5 weeks following the appearance of skin changes. The presence of bacteria, viruses and parasitcs was not detected during the above mentioned examination. Two months after the hospitalization and treatment with the peroral biopsy of small bowel, histopathological changes were not typical for the disease. In histopathological examination villous of small bowel were atrophic, short, flat and cells infiltration in Illama propria (plasmocytes and lymphocytes) Histopathological changes on the small bowel disappeared after the treatment with zinc they disappeared. (Figure 5)

Histopathological changes on the skin are not specific for the disease. Microscopic examination of the skin changes showed scattered parakeratosis and upper epidermal clear cell changes with mild dermal infiltration of lymphocytes. Irregular acanthosis, mild parakeratosis and dermal perivascular infiltration were also observed on another skin section (Figure 6)

Laboratory findings: red blood cell count was 3.3x10^12/L, haemoglobin values 88 g/L, hematokrit level 32%, white blood cell count 8.8x10^9/L, serum iron 7.7 µmol/L. Peripherial blood smear revealed: microcytosis, anulocytosis, hypochromic anemia, anisocytosis (Figure 7). Zinc concentration in plasma was decreased (7.5 µmol/L), total protein value 55 g/L with the decreased daily doses continuously. Skin changes started to become less visible 10 days following the treatment with zinc, hair started to grow after one month, whereas all clinical manifestations of disease disappeared after two months.

5. DISCUSSION

This is the first case with Acrodermatitis enteropathica diagnosed in Kosovo. Global incidence rate is 1:500.000
among the newborns, regardless the gender and ethnic groups (2). Clinical manifestations include the above mentioned skin changes (periocular areas, joints and acral parts of the limbs, inguinal and perianal area), diarrhoea, alopecia, growth failure etc (2, 11, 14). Clinical manifestations of disease at the patient appeared after the ablation and are identical with the cases presented by other authors (2, 8, 17). It is deemed that the breast milk contains zinc factor which is produced in pancreas and which connects zinc and transports it to the epithelial cells of the small bowel. Among the first clinical manifestation in the presented case were changes in the skin and mucocutaneous junction in symmetric manner at the above mentioned regions. Identical changes on skin were described by some other authors in their cases (4, 6, 15), however, it still remains unknown in which part of the body they occurred first. In the case of our patient, the first skin changes appeared on the periocular areas and acnes.

Chronic diarrhoea appeared after two weeks causing the growth failure and sideropenic anemia. Other authors (2, 4, 10, 18) determined that diarrhoea appeared in different time period from 2-4 weeks following the skin changes, whereas growth failure and sideropenic anemia were not present in some cases due to preventive measures. Diarrhoea appeared due to the secondary lactose intolerance, which could not to be hydrolyzed in glucose and galactose under the influence of enzyme lactazta, which lacks due to the histopathological changes of the epithelium of small bowel. Diarrhoea was treated with milk formula without lactose and vegetable diet. Our patient suffered from total alopecia, taking into consideration the fact that zinc is oligolelement important for growth, reproduction and consists of 70 different enzymes. All described cases suffered from total alopecia (4, 6, 7, 12). Histopathological changes were not typical for the disease in the small bowel biopsy which was done before the initial treatment, because other diseases also may appear with the similar histopathological changes (celiac disease, severe malnutrition, giardiasis etc.). These changes disappeared upon the treatment with zinc. Other authors also reported identical changes (1, 10, 13) with their cases with Acrodermatitis enteropathica.

The following histopathological changes were evidenced in the skin biopsy at our patient: parakeratosis, acanthosis and dermal infiltration with lymphocytes. These changes are not typical for the disease. Other authors also reported on having encountered these identical changes in their cases (7, 10). Zinc concentration in plasma was determined during the exacerbation and remission of disease. During the period of the clinical manifestations, zinc concentration was low -7.7 µmol/L (referential values 10.5-22.9 µmol/L). Zinc concentration in plasma was brought into normal in our case one month after continuous treatment with zinc.

Conflict of interest: none declared.

REFERENCES