CASE REPORT

MISMANAGEMENT OF PHENYLKETONURIA: AN UNDERLYING CAUSE OF KWASHIORKOR

Abstract
Objective
Too much restriction of dietary proteins can cause severe protein malnutrition, which can occur in adjusting the diet for some kinds of aminoacidopathies, urea cycle disorder and organic academia. This report presents the case of a 1.5-year-old boy with history of phenylketonuria with a three weeks history of erythematous scaly plaques and edema of his extremities; he had a history of similar skin manifestations three months earlier that resolved spontaneously. The patient had been on very restricted phenylalanine diet. Diagnosed with Kwashiorkor, a phenylalanine level of 0.4 mg/dl, the child was hospitalized and put on a special diet and given the appropriate antibiotic; after a few days of treatment his condition improved.
We underscore the importance of education for those considering prescription of diet restriction and emphasize the regulation of balanced diet in such patients.

Key words: Phenylketonuria, kwashiorkor, protein malnutrition

Introduction
Phenylketonuria (PKU) is an inborn error of protein metabolism that results from an impaired ability to metabolize the essential amino acid phenylalanine. The term kwashiorkor means “sickness of weaning”. It refers to an inadequate protein intake with reasonable caloric (energy) intake. Edema is characteristic of kwashiorkor but is absent in marasmus (1).

Case report
A 1.5 year-old boy presented with a three week-history of erythematous scaly plaques on his extremities, diaper-region, and face. Three months later similar lesions appeared over much of his body, but resolved spontaneously; the patient developed cracked lips, mucosal ulcers, thinning of the hair, and 2 plus non pitting edema on both extremities, two weeks after the skin lesions (figure1). He was a confirmed case of phenylketonuria since the 11th month of his life, and had hence been on a diet, fed only by analogue X P formula; phenylalanine was omitted totally. His medications include clonazepam and Phenobarbital for seizure disorder. He had also been prescribed folic acid and ferrogluboline.
The child was a full term infant, born without complications. While his past medical history was significant for neurodevelopmental delay and visual inattention, his older brother was and is healthy. On examination, the patient was irritable and diffusely edematous and had a “chubby” appearance. His skin manifestations
include multiple annular plaques with hyperkeratotic border and desquamated centers and were confluent over the extremities, buttocK, and face (figure1); in addition he has angular chelitides. Neurological exam revealed poor head control, microcephaly, and poor eye contact. Laboratory studies revealed a hemoglobin of 6.1 and hematocrite 18.3, white blood cell count 18200(poly: 59%) and platlate count 285000. total protein and serum albumin were 4.9 mg/dL and 2.1mg/dL, respectively. BS, BUN, Cr, Ca, Ph, and tranaminas were in normal ranges. Phenylalanine level was 0.4 mg/dl. The child was hospitalized and put on a special diet, multivitamins, zinc, folate, albumin, and appropriate antibiotic. The skin lesions improved dramatically using a combination of Mupirocine, Hydrocortisone, and Clotrimazol; oral lesions responded to Nystatin and Metronidazole.

Discussion
Although the “flaky paint” dermatosis manifested in this child is a classic feature of Kwashiorkor, the disease is known to present in a variety of different ways. Many researchers of kwashiorkor have documented the role of other nutritional deficiency, including riboflavin, niacin, B6, and B12, as an etiologic factor for the lack of appearance of clinical signs and symptoms (2). Most probably because it found almost only in protein–rich foods, riboflavin deficiency is very common in kwashiorkor patients, and may hence account for the prevalence of angular ostomatitis and angular cheilitis in kwashiorkor (3, 4). Another deficiency that may be confused with or overlap kwashiorkor is essential free fatty acid (EFA), that manifests as xerosis of the skin, dry thickened, erythematous desquamating plaques, and alopecia with hypopigmentation of the remaining hair (4, 5-8). Deliberately restricted dietary intake may induce protein energy malnutrition including anorexia nervosa, lactate intolerance, and urea cycle and related metabolic disorders, phenylketonuria, hereditary tyrosinemia, and organic aciduria (2). In our case, highly restricted phenylalanine diet resulted in signs and symptoms indicating kwashiorkor, with his plasma phenylalanine levels being 0.4 mg/dL on admission; Plasma phenylalanin concentrations of normal infants range from 2 to 6 mg/dL. Such low concentrations of
References


Figures 1(a-c): The child’s buttocks and upper extremities show the desquamative rash with hyperpigmented borders “flaky-paint” dermatosis