

Clinical case blog

Title: Tyrosinemia Type 2 Presented as Food Allergy

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7 year-old-girl referred to the pediatric gastroenterology clinic as a case of food allergy, due to painful plantopalmar lesions, erupting after eating meat, poultry and dairy products. The child was unable to stand due to severe pain. The lesions were symmetrical, patchy, hyperkeratotic and painful. Lesions were suggestive of Tyrosinemia type 2. Her neurological and ophthalmological evaluation showed no abnormality. Elevated serum tyrosine level confirmed the diagnosis. After starting low protein diet and low phenylalanine and tyrosine formula, her symptoms improved.

Tyrosinemia type 2 (Richner-Hanhart Syndrome) is an autosomal recessive disorder [1]. The disease is caused by a deficiency of the hepatic enzyme tyrosine aminotransferase (TAT), leading to elevated levels of tyrosine in blood and urine. Skin and eyes are the cardinal organs involved. Nervous system involvement is well documented also.

Typical skin lesions are painful, well-demarcated hyperkeratosis on the palms and soles. Lesions may begin as bullae and erosions that progress to crusted plaques [2]. Ocular manifestations include corneal clouding, opacities, scars, ulcers or neovascularizations [2]. Skin and ocular manifestations are caused by inflammatory response to intracellular accumulation of Tyrosine crystals [3]. The relationship between the amount of protein intake and flaring of ocular symptoms is well-established [4]. This might explain symptoms worsening associated with specific protein intake in our case, which lead to misinterpret the relation as allergy.

Restricting tyrosine and phenylalanine containing food can reverse eye and skin manifestations and prevent neurological involvement [5]. Tyrosinemia type 2 should be suspected in patients with palmoplantar keratosis. Worsening of the lesions with protein diet should not be mistaken for food allergy.

Figure: Symmetrical patchy hyperkeratotic yellow plaques on the weight-bearing areas of both feet. Patients refuse to walk due to severe pain

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