Corneal Ulceration and Acrodermatitis Dysmetabolica During Treatment of a Patient with Maple Syrup Urine Disease

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Introduction: Maple syrup urine disease (MSUD) is an autosomal recessive disease which is an inborn error of branched chain amino acids (BCAAs). We report an 15 month old boy with MSUD who developed an episode of cutaneous lesions and deepithelialization of the cornea as a result of isoleucine deficiency.

Case presentation: A 15 month old boy with MSUD who does not go to regular outpatient clinic controls was presented with eczematous desquamation on hands, feet and gluteal regions and redness of the eyes. Acrodermatitis dysmetabolica due to isoleucine deficiency was made based on the clinical findings and low isoleucine levels. Ophthalmological examination revealed spontaneous corneal ulcerations due to total deepithelialization of the cornea in the left eye. He was treated with eye-ointment (Moxifloxacin") bilaterally, zinc and isoleucine (10 0 mg/day) supplementation. In addition, skin care with emollients was administered. The patient showed a rapid improvement in the skin lesions which started to recover within 2 days and healed in 10 days with no hyperpigmentation. Cornea transplantation did not recommended by ophthalmologists.

Discussion: Acrodermatitis dysmetabolica and corneal ulceration in MSUD patients could arise from multiple causes. Clinical status and BCAA levels, should be closely monitored in MSUD patients.