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Clinical Image Open Access

Maple Syrup Urine Disease

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Clinical Image

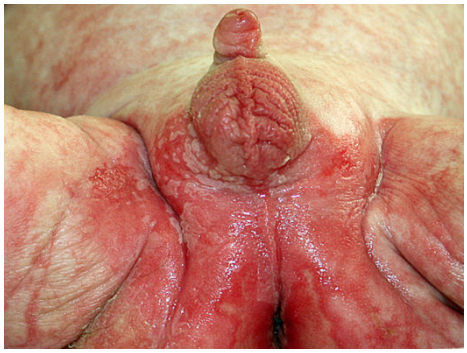

Figure 1: Physical examination of maple syrup urine disease.

A 9 day-old male infant presented with generalized seizures, irritability, lethargy, vomiting, skin abrasions in the genital area and sweet-smelling urine with a resemblance to burnt sugar in odour. Physical examination showed cutis marmorata in abdomen and thorax, swelling of external genitalia associated with skin abrasions as well as lacerations in perineum and upper medial side of both thighs, and generalized erythema (Figure 1). Plasma and urine amino acid analysis revealed a perceptible increase of branched chain amino acids (BCAA). The diagnosis of maple syrup urine disease was confirmed on low branched-chain-α-keto acid dehydrogenase complex activity in lymphocytes. The patient was treated with a BCAA-free diet and thiamine (0.2 milligrams daily), with a good response.