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P10 Treatment-refractory psoriasiform dermatitis resulting from a rare genetic alteration in MSMO1 with marked improvement with combined cholesterol and statin use

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Abstract

An infant of 19 months presented to dermatology with a widespread psoriasiform rash favouring inguinal and neck flexures. There was a known family history of psoriasis but no other concerns reported. This progressed to become erythrodermic despite multiple topical treatments as well as systemic treatments including ciclosporin and zinc supplements. Skin biopsy suggested psoriasis or nutritional deficiency. Oral acitretin reduced skin severity significantly. However, there was continued impact from areas of inflammatory skin with only partial response to topical treatments.

Other syndromic features were apparent from presentation and developed over follow-up with global developmental delay, microcephaly, congenital cataracts corrected age 3, and skeletal abnormalities (inward curving little finger).

A genetic defect in an enzyme in the cholesterol pathway was subsequently confirmed with alteration (compound heterozygous) in *MSM01* gene which code for methylsterol-monoxygenase-1enzyme. Deficiency in this key player in cholesterol synthesis causes downstream deficiencies in hormones and other compounds required for healthy development and functioning. It also causes an upstream accumulation of toxic methylsterols. This results in this rare metabolic condition with neurodevelopmental, dermatological, ocular and skeletal manifestations.

Topical *(2% simvastatin/2% cholesterol cream)* and oral statin was commenced to reduce the toxic accumulation of methylsterols, along with cholesterol replacement to replace the downstream deficiency. Improvement in her skin, with almost complete clearance after only after a few months of treatment has been life changing.

Awareness of potential genetic metabolic causes of childhood skin conditions is key to ensure appropriate support and early effective treatments.