Netherton's syndrome

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History. After an uncomplicated pregnancy the boy was born as the first child of healthy Turkish consanguineous parents. Immediately after birth a red and scaly skin was noted. By 2 months the child was transferred to the Sophia Children's Hospital (SCH).

Examination. At birth a red skin was noted with peeling on the chin, arms and hands. He did not have the appearance of a collodion baby. One week after birth there was gross lamellar scaling. At the age of 2 months the skin was yellow without any subcutaneous fat. There were fissures but no bullae. The nails were normal and the hair was sparse. There were 'dermogenetic' contractures.

Histology and electron microscopy. The epidermis showed parakeratosis and acanthosis. The granular layer was virtually absent. There was considerable oedema in the epidermis. On electron microscopy desmosomes were irregularly developed and locally absent. There were small numbers of tonofilaments and intercellular oedema (assessment with H.Traupe, Münster, now Nijmegen).

Additional investigations. Hair root examination: 20% anagen, 80% catagen ($n=25$ hairs). Light microscopy of hairs revealed variable hair-shaft diameter but generally the shafts were thin (10–20 μm). Features of trichorrhexis invaginata (bamboo hair) were present. On polarization microscopy there were no special additional abnormalities.

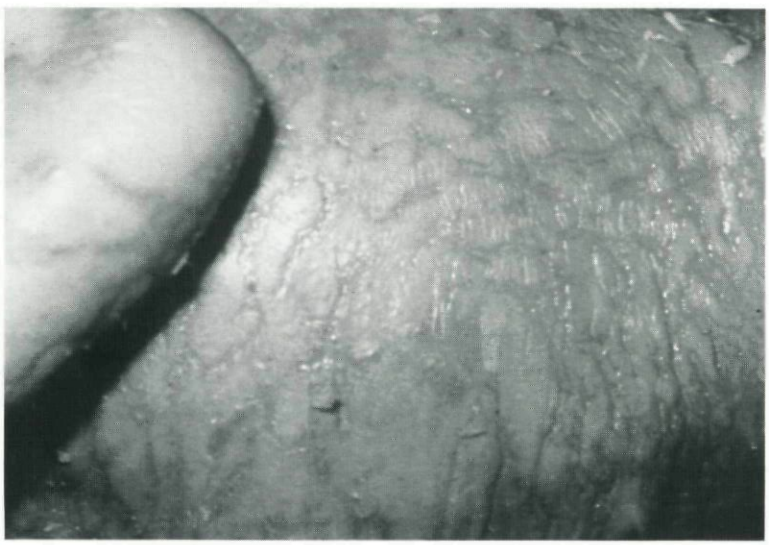

FIGURE 1. Netherton's syndrome.

Course. Recurrent severe infections, a failure to thrive, feeding problems, severe metabolic acidosis, recurrent anaemia, liver failure and dehydration (fluid/protein leakage through the skin) and disturbed thermoregulation were encountered during the course of hospitalization. The child later died.

Comment. Netherton's syndrome is a disorder of keratinization with a non-bullous ichthyosiform erythroderma that often has a fatal outcome within the first few months of life. Typical features are ichthyosiform desquamation with a double collarette (ichthyosis linearis circumflexa), bamboo hair and often an atopic diathesis. It is believed to be an autosomal recessive disorder with considerable penetration in females.

REFERENCE

1 Anton Lamprecht I. Netherton syndrome. *Hum Genet* 1985; **71**: 301–11.