Opis choroby *

Definicja

A rare genetic disease characterized by abnormalities in renal ion transport, ectodermal gland homeostasis, and epidermal integrity, resulting in generalized hypohidrosis, heat intolerance, salt-losing nephropathy, electrolyte imbalance, lacrimal gland dysfunction, ichthyosis, and xerostomia. Development of nephrolithiasis and severe enamel wear have also been described. Laboratory findings include hypermagnesemia, hypokalemia, hypercalcemia, and hypocalciuria.

Dane

Klasyfikacja
Choroba

Synonimy
HELIX syndrome
Zespół HELIX

Kod ORPHA
528105

Kod OMIM
617671

Kod ICD10
N25.8

Kod ICD11
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*Źródło

orphanet