DISEASE:
Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome

NAME:	Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome
DESCRIPTION:	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.
ORPHACODE:	528105
SYNONYMS:	HELIX syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	CLDN10
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RELATED CONTENT

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- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

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