DISEASE:
Ichthyosis-hypotrichosis syndrome

NAME:	Ichthyosis-hypotrichosis syndrome
DESCRIPTION:	Ichthyosis-hypotrichosis syndrome is characterised by congenital ichthyosis and hypotrichosis. It has been described in three members of a consanguineous Arab Israeli family. The syndrome is transmitted as an autosomal recessive trait and is caused by a missense mutation in the ST14 gene, encoding the recently identified protease, matriptase. Analysis of skin samples from the patients suggests that this enzyme plays a role in epidermal desquamation.
ORPHACODE:	91132
SYNONYMS:	Hypotrichosis-congenital ichthyosis syndrome IFAH syndrome IHS Ichthyosis-follicular atrophoderma-hypotrichosis syndrome Ichthyosis-follicular atrophoderma-hypotrichosis-hypohidrosis syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ST14
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