Ichthyosis-hypotrichosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ichthyosis-hypotrichosis syndrome. ORPHA:91132

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.