Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hypohidrotic ectodermal dysplasia-hypothyroidism-ciliary dyskinesia syndrome. ORPHA:1882

A rare, genetic, ectodermal dysplasia syndrome characterized by the association of hypohidrotic ectodermal dysplasia (manifesting with the triad of hypohidrosis, anodontia/hypodontia and hypotrichosis) with primary hypothyroidism and respiratory tract ciliary dyskinesia. Patients frequently present urticaria pigmentosa-like skin pigmentation, increased mast cells and melanin depositions in the dermis and severe, recurrent chest infections. There have been no further descriptions in the literature since 1986.