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Sparse hair-short stature-skin anomalies syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Sparse hair-short stature-skin anomalies syndrome. ORPHA:79132*

Sparse hair-short stature-skin anomalies syndrome combines short stature, sparse hair, skin hyperpigmentation and urticaria-like reactions on the hands and arms. An upper central incisor, hypoplastic thumbs and/or palmoplantar hyperkeratosis may also be present. It is thought to be a rare form of ectodermal dysplasia and has been described at least once in a mother and her three sons. Transmission is autosomal dominant, or X-linked.