Dermatoleukodystrophy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Dermatoleukodystrophy. ORPHA:1659

Dermatoleukodystrophy is characterised by the association of a progressive leukodystrophy marked by generalised mental and motor impairment with the presence of thickened and wrinkled skin. It has been described in a Japanese brother and sister born to healthy parents. Both patients died in early childhood.