Skin fragility-woolly hair-palmoplantar keratoderma syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Skin fragility-woolly hair-palmoplantar keratoderma syndrome. ORPHA:293165

Skin fragility-woolly hair-palmoplantar keratoderma syndrome is a rare, genetic, ectodermal dysplasia syndrome characterized by persistent skin fragility which manifests with blistering and erosions due to minimal trauma, woolly hair with variable alopecia, hyperkeratotic nail dysplasia, diffuse or focal palmoplantar keratoderma with painful fissuring, and no cardiac abnormalities. Perioral hyperkeratosis may also be associated.