Parana hard skin syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Parana hard skin syndrome. ORPHA:2812

Parana hard skin syndrome is a rare genetic skin disorder characterized by very early-onset of progressive skin thickening over the entire body (except for eyelids, neck and ears), progressively limited joint mobility with gradual freezing of joints, and eventual severe chest and abdomen movement restriction, manifesting with restrictive pulmonary disease, which may lead to death. Additional features include severe growth restriction and osteoporosis. There have been no further descriptions in the literature since 1974.