Harlequin ichthyosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Harlequin ichthyosis. ORPHA:457

Harlequin ichthyosis (HI) is the most severe variant of autosomal recessive congenital ichthyosis (ARCI; see this term). It is characterized at birth by the presence of large, thick, plate-like scales over the whole body associated with severe ectropion, eclabium, and flattened ears, that later develops into a severe scaling erythroderma.