Junctional epidermolysis bullosa with respiratory and renal involvement

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Junctional epidermolysis bullosa with respiratory and renal involvement. ORPHA:306504

Congenital nephrotic syndrome-interstitial lung disease-epidermolysis bullosa syndrome is a life-threatening multiorgan disorder which develops in the first months of life, presenting with respiratory distress and proteinuria in the nephrotic range, and leading to severe interstitial lung disease and renal failure. Some patients additionally display cutaneous alterations, ranging from blistering and skin erosions to an epidermolysis bullosa-like phenotype, with toe nail dystrophy and sparse hair.