Opis choroby *

Definicja

A rare, genetic, renal disease characterized by hereditary nephritis leading to nephrotic syndrome and end-stage renal failure associated with sensorineural hearing loss and pretibial skin blistering followed by atrophy. Other reported manifestations include bilateral lacrimal duct stenosis, dystrophic teeth and nails, bilateral cervical ribs, unilateral kidney, distal vaginal agenesis and anemia due to beta-thalassemia minor.

Dane

Klasyfikacja
Choroba

Synonimy
EBS with nephropathy
Zespół zespołu nefrotycznego, utraty słuchu, przedpiszczelowego pęcherzowego oddzielania się naskórka
Epidermolysis bullosa simplex with nephropathy
Nephrotic syndrome-hearing loss-epidermolysis bullosa syndrome

Kod ORPHA
300333

Kod OMIM
609057

Kod ICD10
N08.2

Kod ICD11
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*Źródło

orphanet