

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, przedpyszczelowego 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](#)

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