DISEASE:
Nephrotic syndrome-epidermolysis bullosa-sensorineural deafness syndrome

NAME:	Nephrotic syndrome-epidermolysis bullosa-sensorineural deafness syndrome
DESCRIPTION:	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.
ORPHACODE:	300333
SYNONYMS:	EBS with nephropathy Epidermolysis bullosa simplex with nephropathy Nephrotic syndrome-hearing loss-epidermolysis bullosa syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	CD151
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RELATED CONTENT

Related Genetic Tests

- Epidermolysis bullosa (gene panel)
- Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- CD151 molecule (Raph blood group)

Related Gene Panels

- Epidermolysis bullosa and bladder diseases (60 genes) - KUL
- Nephrotic syndrome, FSGS, Alport syndrome (76 genes) - IPG