DISEASE:
Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome

NAME:	Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome
DESCRIPTION:	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.
ORPHACODE:	306504
SYNONYMS:	ILNEB syndrome JEB with interstitial lung disease and nephrotic syndrome Junctional epidermolysis bullosa with interstitial lung disease and nephrotic syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ITGA3
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RELATED CONTENT

Related Genetic Tests

- Child Interstitial Lung Disease (child - gene panel)
- Epidermolysis bullosa (gene panel)
- Pulmonary Fibrosis (gene panel) + rs35705950 of MUC5B gene

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- integrin subunit alpha 3

Related Gene Panels

- Epidermolysis bullosa and bladder diseases (60 genes) - KUL
- Pulmonary Fibrosis (21 genes) + rs35705950 (MUC5B gene) - KUL
- chILD (34 genes) - KUL