DISEASE:
Interstitial lung disease due to ABCA3 deficiency

NAME:	Interstitial lung disease due to ABCA3 deficiency
DESCRIPTION:	Interstitial lung disease due to ABCA3 deficiency is a rare genetic respiratory disease characterized by a variable clinical outcome ranging from a fatal respiratory distress syndrome in the neonatal period to chronic interstitial lung disease developing in infancy or childhood with chronic cough, rapid breathing, shortness of breath and recurrent pulmonary infections. Clinical manifestations of respiratory failure include grunting, intercostal retractions, nasal flaring, cyanosis, and progressive dyspnea.
ORPHACODE:	440402
SYNONYMS:	Interstitial lung disease due to ATP-binding cassette subfamily A member 3 deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ABCA3
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

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- Pulmonary Fibrosis (gene panel) + rs35705950 of MUC5B gene

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- ATP binding cassette subfamily A member 3

Related Gene Panels

- Pulmonary Fibrosis (21 genes) + rs35705950 (MUC5B gene) - KUL
- chILD (34 genes) - KUL