DISEASE:
Interstitial lung disease due to SP-C deficiency

NAME:	Interstitial lung disease due to SP-C deficiency
DESCRIPTION:	A rare genetic interstitial lung disease characterized by diffuse lung disease of variable phenotype ranging from severe respiratory insufficiency in infancy to asymptomatic adults, due to surfactant protein C deficiency. Typical presentation in infancy includes dyspnea, cough, wheezing, and gradual cyanosis, with or without failure to thrive. Radiological findings include diffuse ground-glass opacities in neonates, later interstitial thickening associated with lung hyperinflation, intraparenchymal/subpleural cysts, honeycombing, subpleural nodules, or bronchiectasis. Infiltrates and air leaks are frequent complications.
ORPHACODE:	440392
SYNONYMS:	Interstitial lung disease due to surfactant protein C deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SFTPC
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RELATED CONTENT

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- Centrum Menselijke Erfelijkheid - KUL

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