# DISEASE:
# Neonatal acute respiratory distress due to SP-B deficiency

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| **NAME:** | Neonatal acute respiratory distress due to SP-B deficiency |
| **DESCRIPTION:** | A rare genetic interstitial lung disease characterized by progressive, life-threatening, refractory respiratory distress in full-term neonates associated with surfactant protein B deficiency. In most cases, the disease is fatal within the first months of life. Lung biopsy reveals changes characteristic of pulmonary alveolar proteinosis with interstitial fibrosis and inflammation, as well as accumulation of lipid-rich, eosinophilic, proteinaceous, granular material consisting of desquamated type II pneumocytes and foamy macrophages within the alveolar air spaces. |
| **ORPHACODE:** | 217563 |
| **SYNONYMS:** | Neonatal acute respiratory distress due to surfactant protein B deficiency |
| **XREF(S):** | Orphanet<br>ICD-10<br>OMIM |
| **ANALYTE(S):** | SFTPB |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

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

## Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- surfactant protein B

## Related Gene Panels

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