

Full name:	chILD (34 genes) - KUL
Version number:	chILD_v3
Laboratory:	<u>Centrum Menselijke Erfelijkheid - KUL</u>
Created:	15 Dec 2020 - 17:27
Changed:	26 Jul 2023 - 10:25

Related Diseases

- Autoimmune interstitial lung disease-arthritis syndrome
- Chronic respiratory distress with surfactant metabolism deficiency
- Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome
- Infant acute respiratory distress syndrome
- Infantile-onset pulmonary alveolar proteinosis-hypogammaglobulinemia
- Interstitial lung disease due to ABCA3 deficiency
- Interstitial lung disease due to SP-C deficiency
- Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome
- Neonatal acute respiratory distress due to SP-B deficiency
- Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome
- Pulmonary alveolar microlithiasis
- Spinal muscular atrophy with respiratory distress type 1
- Spinal muscular atrophy with respiratory distress type 2

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ABCA3</u>	95.00	0	NM_001089.3 / interpretable range CS1>95%
<u>AP3B1</u>	95.00	0	NM_003664.5/ interpretable range CS1>95%
<u>AP3D1</u>	95.00	0	NM_001261826.3/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>BLOC1S3</u>	95.00	0	NM_212550.5/ interpretable range CS1>95%
<u>BLOC1S5</u>	95.00	0	NM_201280.3/ interpretable range CS1>95%
<u>BLOC1S6</u>	95.00	0	NM_012388.4/ interpretable range CS1>95%
<u>COPA</u>	95.00	0	NM_004371.4/ interpretable range CS1>95%
<u>CSF2RA</u>	95.00	0	NM_006140.6/ interpretable range CS1>95%
<u>CSF2RB</u>	95.00	0	NM_000395.3/ interpretable range CS1>95%
<u>DTNBP1</u>	95.00	0	NM_032122.5/ interpretable range CS1>95%
<u>FLNA</u>	95.00	0	NM_001456.4/ interpretable range CS1>95%
<u>FOXF1</u>	95.00	0	NM_001451.3/ interpretable range CS1>95%
<u>GATA2</u>	95.00	0	NM_032638.5/ interpretable range CS1>95%
<u>HPS1</u>	95.00	0	NM_000195.5/ interpretable range CS1>95%
<u>HPS3</u>	95.00	0	NM_022081.6/ interpretable range CS1>95%
<u>HPS4</u>	95.00	0	NM_022081.6/ interpretable range CS1>95%
<u>HPS5</u>	95.00	0	NM_181507.2/ interpretable range CS1>95%
<u>HPS6</u>	95.00	0	NM_024747.6/ interpretable range CS1>95%
<u>MARS1</u>	95.00	0	NM_004990.4/ interpretable range CS1>95%
<u>MUC5B</u>	95.00	0	NM_002458.3/ interpretable range CS1>95%
<u>NKX2-1</u>	95.00	0	NM_001079668.3/ interpretable range CS1>95%
<u>PARN</u>	95.00	0	NM_002582.4/ interpretable range CS1>95%

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>RAB5B</u>	95.00	0	NM_002868.4/ interpretable range CS1>95%
<u>RTEL1</u>	95.00	0	NM_032957.5/ interpretable range CS1>95%
<u>SFTPA1</u>	95.00	0	NM_005411.5/ interpretable range CS1>95%
<u>SFTPA2</u>	95.00	0	NM_001098668.4/ interpretable range CS1>95%
<u>SFTPB</u>	95.00	0	NM_198843.3/ interpretable range CS1>95%
<u>SFTPC</u>	95.00	0	NM_003018.4/ interpretable range CS1>95%
<u>SLC34A2</u>	95.00	0	NM_006424.3/ interpretable range CS1>95%
<u>SLC7A7</u>	95.00	0	NM_001126106.4/ interpretable range CS1>95%
<u>STING1</u>	95.00	0	NM_198282.4/ interpretable range CS1>95%
<u>TBX4</u>	95.00	0	NM_018488.3/ interpretable range CS1>95%
<u>TERC</u>	95.00	0	NR_001566.1/ interpretable range CS1>95%
<u>TERT</u>	95.00	0	NM_198253.3/ interpretable range CS1>95%