DISEASE:
Spinal muscular atrophy with respiratory distress type 2

NAME:	Spinal muscular atrophy with respiratory distress type 2
DESCRIPTION:	Spinal muscular atrophy with respiratory distress type 2 is a rare, genetic, motor neuron disease characterized by progressive early respiratory failure associated with diaphragm paralysis, distal muscular weakness, joint contractures, and axial hypotonia with preserved antigravity limb movements. Phenotype overlaps considerably with SMARD type 1 but is differentiated by a mutation in a different gene.
ORPHACODE:	404521
SYNONYMS:	Diaphragmatic spinal muscular atrophy type 2 SMARD2 Severe infantile axonal neuropathy with respiratory failure type 2 X-linked spinal muscular atrophy with respiratory distress
XREF(S):	Orphanet
ANALYTE(S):	LAS1L
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- Centrum Medische Genetica - UZ Brussel VUB
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