Spinal muscular atrophy with respiratory distress type 1

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Spinal muscular atrophy with respiratory distress type 1. ORPHA:98920

Spinal muscular atrophy with respiratory distress type 1 is a rare genetic motor neuron disease characterized by severe respiratory distress/respiratory failure in association with diaphragmatic eventration and palsy, as well as progressive, symmetrical, distal-to-proximal muscle weakness and atrophy (in lower limbs especially). Patients typically have a history of intrauterine growth retardation, low birth weight, feeble cry, weak suck and failure to thrive and present with inspiratory stridor, recurrent episodes of dyspnea or apnea, cyanosis and absent deep tendon reflexes. Kyphosis/scoliosis, foot deformities and joint contractures are frequently associated features.