Infantile-onset X-linked spinal muscular atrophy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Infantile-onset X-linked spinal muscular atrophy. ORPHA:1145

X-linked distal arthrogryposis multiplex congenital (SMAX2) is a rare form of spinal muscular atrophy characterized by the neonatal onset of severe hypotonia, areflexia, profound weakness, multiple congenital contractures, facial dysmorphic features (myopathic face with open, tent-shaped mouth), cryptorchidism, and mild skeletal abnormalities (i.e. kyphosis, scoliosis), that is often preceded by polyhydramnios and reduced fetal movements in utero and followed by bone fractures shortly after birth. SMAX2 patients often have a limited life span, often succumbing to the disease within 2 years, as muscle weakness is progressive and chest muscle involvement eventually leads to ventilatory insufficiency and respiratory failure.