Fatal infantile hypertonic myofibrillar myopathy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Fatal infantile hypertonic myofibrillar myopathy. ORPHA:280553

Fatal infantile hypertonic myofibrillar myopathy is a rare, genetic skeletal muscle disease characterized by muscle stiffness and rigidity, hypertonia, weakness, respiratory distress and normal cognition. Patients have persistently elevated creatine kinase and histopathology is typical of myofibrillar myopathy. The manifestation onset follows the short period of normal infantile development and leads to progressive respiratory insufficiency and early death.