Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome. ORPHA:478049

Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome is rare, genetic, neurometabolic disease characterized by global developmental delay, severe hypotonia, seizures, cataracts, cardiomyopathy (including left or bi-ventricular hypertrophy, dilated cardiomyopathy) and left ventricular non-compaction, typically resulting in infantile or early-childhood death. Patients usually present metabolic lactic acidosis, failure to thrive, head lag, respiratory problems and decrease in respiratory chain complex activity. Highly variable cerebral abnormalities have been reported and include microcephaly, prominent extra-axial cerebrospinal fluid spaces, diffuse neuronal loss and cortical/white matter gliosis.