Combined oxidative phosphorylation defect type 25

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Combined oxidative phosphorylation defect type 25. ORPHA:447954

Combined oxidative phosphorylation defect type 25 is a rare mitochondrial oxidative phosphorylation disorder with decreased respiratory complex I and IV enzyme activities, characterized by hypotonia, global developmental delay, neonatal onset of progressive pectus carinatum without other skeletal abnormalities, poor growth, sensorineural hearing loss, dysmorphic features and brain abnormalities such as cerebral atrophy, quadriventricular dilatation and thin corpus callosum posteriorly.