# Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3. ORPHA:168566*

Combined oxidative phosphorylation deficiency type 3 is an extremely rare clinically heterogenous disorder described in about 5 patients to date. Clinical signs included hypotonia, lactic acidosis, and hepatic insufficiency, with progressive encephalomyopathy or hypertrophic cardiomyopathy.