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Severe X-linked mitochondrial encephalomyopathy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Severe X-linked mitochondrial encephalomyopathy. ORPHA:238329

Severe X-linked mitochondrial encephalomyopathy is an extremely rare mitochondrial respiratory chain disease resulting in a neurodegenerative disorder characterized by psychomotor delay, hypotonia, areflexia, muscle weakness and wasting in the two patients reported to date.