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Severe neurodegenerative syndrome with lipodystrophy

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Severe neurodegenerative syndrome with lipodystrophy. ORPHA:363400*

Severe neurodegenerative syndrome with lipodystrophy is a rare, genetic, neurodegenerative disorder characterized by progressive psychomotor and cognitive regression (manifesting with gait ataxia, spasticity, loss of language, mild to severe intellectual disability, pyramidal and extrapyramidal signs and, frequently, development of tetraplegia or tetraparesis) associated with variable degrees of lipodystrophy, hepatomegaly, hypertriglyceridemia and muscular hypertrophy. Hyperactivity, tremor and development of seizures may also be associated.