DISEASE:
Severe neurodegenerative syndrome with lipodystrophy

NAME:	Severe neurodegenerative syndrome with lipodystrophy
DESCRIPTION:	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 tretraplegia or tetraparesis) associated with variable degrees of lipodystrophy, hepatomegaly, hypertriglyceridemia and muscular hypertrophy. Hyperactivity, tremor and development of seizures may also be associated.
ORPHACODE:	363400
SYNONYMS:	Severe neurodegenerative syndrome due to BSCL2 deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	BSCL2
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RELATED CONTENT

Related Genetic Tests

- Congenital generalized lipodystrophy type 2 / Spastic paraplegia-17 / Hereditary motor neuronopathy type VA / Silver spastic paraplegia syndrome (hot spot mutation - p.Asn88Ser; p.Ser90; p.Arg96His)
- Lipodystrophy (2 genes)
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)
- Neuropathy (gene panel)
- Peripheral neuropathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- BSCL2 lipid droplet biogenesis associated, seipin

Related Gene Panels

- Lipodystrophy (2 genes) - IPG
- Neuromuscular disorders (166 genes) - VUB

- Neuropathy (148 genes) - IPG
- Neuropathy (genepanel) - UZA

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