DISEASE:
Congenital generalized lipodystrophy

NAME:	Congenital generalized lipodystrophy
DESCRIPTION:	A rare autosomal recessive form of lipodystrophy characterized by the association of generalized lipoatrophy with acromegaloid features, muscle hypertrophy, insulin resistance, hypertriglyceridemia, and liver steatosis.
ORPHACODE:	528
SYNONYMS:	BSCL Berardinelli-Seip congenital lipodystrophy Berardinelli-Seip syndrome CGL Lipoatrophic diabetes
XREF(S):	Orphanet OMIM MedDRA ICD-10 OMIM OMIM OMIM OMIM

ANALYTE(S):	CAVIN1 PPARG BSCL2 AGPAT2 CAV1 FOS
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RELATED CONTENT

Related Genetic Tests

- Congenital generalized lipodystrophy type 1
- 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)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- 1-acylglycerol-3-phosphate O-acyltransferase 2
- BSCL2 lipid droplet biogenesis associated, seipin
- caveolin 1
- caveolae associated protein 1
- Fos proto-oncogene, AP-1 transcription factor subunit
- peroxisome proliferator activated receptor gamma

Related Gene Panels

- Lipodystrophy (2 genes) - IPG

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