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# Berardinelli-Seip Congenital Lipodystrophy

National Cancer Institute

## Source

National Cancer Institute. *Berardinelli-Seip Congenital Lipodystrophy*. NCI Thesaurus. Code C84594.

A genetic disorder caused by mutations in the AGPAT2 and BSCL2 genes. It is characterized by lipoatrophy in the trunk, face, and extremities, hypertriglyceridemia, muscle hypertrophy, cardiomyopathy and hepatomegaly. Patients develop insulin resistance, leading to diabetes mellitus.