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Cholesterol Side-Chain Cleavage Deficiency

National Cancer Institute

Source

National Cancer Institute. *Cholesterol Side-Chain Cleavage Deficiency*. NCI Thesaurus. Code C131422.

CLAH due to loss-of-function mutations in the CYP11A1 gene, resulting in decreased or absent activity of the enzyme P450scc, which leads to reduced conversion of cholesterol to pregnenolone, the first step in steroidogenesis.