Presynaptic Congenital Myasthenic Syndrome 6

National Cancer Institute

Source

National Cancer Institute. Presynaptic Congenital Myasthenic Syndrome 6. NCI Thesaurus. Code C132292.

Congenital myasthenic syndrome caused by mutation(s) in the CHAT gene, encoding choline O-acetyltransferase. It is inherited in an autosomal recessive manner.