

Open Peer Review on Qeios

Presynaptic Congenital Myasthenic Syndrome 6

National Cancer Institute

Source

National Cancer Institute. <u>Presynaptic Congenital Myasthenic Syndrome 6</u>. 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.

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