Open Peer Review on Qeios

Congenital Myasthenic Syndrome 5

National Cancer Institute

Source

National Cancer Institute. <u>Congenital Myasthenic Syndrome 5</u>. NCI Thesaurus. Code C129304.

Congenital myasthenic syndrome caused by mutation(s) in the COLQ gene, encoding acetylcholinesterase collagenic tail peptide. It is inherited in an autosomal recessive manner.