

Open Peer Review on Qeios

Temtamy Syndrome

National Cancer Institute

Source

National Cancer Institute. <u>Temtamy Syndrome</u>. NCI Thesaurus. Code C148371.

An extremely rare autosomal recessive condition caused by mutation(s) in the C12orf57 gene, encoding protein C10. It is characterized by agenesis/hypoplasia of the corpus callosum, associated with developmental delay, and variable craniofacial and skeletal abnormalities.

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