Open Peer Review on Qeios

17q21 Microdeletion Syndrome

National Cancer Institute

Source

National Cancer Institute. 17q21 Microdeletion Syndrome. NCI Thesaurus. Code C75470.

A genetic syndrome caused by microdeletions in chromosome 17q21. The microdeletions encompass the MAPT and CRHR1 genes. It is characterized by mental retardation, hypotonia, distinctive facial features (long face, low-set ears, and pear-shaped nose), friendly behavior, and heart defects.