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Miller-Dieker Syndrome

National Cancer Institute

Source

National Cancer Institute. *Miller-Dieker Syndrome*. NCI Thesaurus. Code C124852.

A rare syndrome caused by deletion of genetic material in the short arm of chromosome 17. It is characterized by an abnormally smooth brain with fewer folds and grooves. It results in intellectual disability, developmental delay, seizures, spasticity, hypotonia, and feeding difficulties. Affected individuals have distinctive facial features that include a prominent forehead, midface hypoplasia, small, upturned nose, low-set ears, small jaw, and thick upper lip.