Open Peer Review on Qeios

Kleefstra Syndrome

National Cancer Institute

Source

National Cancer Institute. <u>Kleefstra Syndrome</u>. NCI Thesaurus. Code C129976.

A condition caused by mutation or deletion of the EHMT1 gene, encoding histone-lysine N-methyltransferase EHMT1. It is characterized by severe intellectual disability, hypotonia, cardiac defects, and characteristic facial features.