Open Peer Review on Qeios

Potocki-Lupski Syndrome

National Cancer Institute

Source

National Cancer Institute. <u>Potocki-Lupski Syndrome</u>, NCI Thesaurus. Code C124846.

A rare genetic disorder characterized by hypotonia, failure to thrive, mental retardation, developmental disorders, congenital anomalies, and autism spectrum disorders. The majority of patients harbor a microduplication of chromosome 17p11.