Joubert Syndrome Type 4

National Cancer Institute

Source

National Cancer Institute. Joubert Syndrome Type 4. NCI Thesaurus. Code C74997.

A rare genetic syndrome caused by mutations in the NPHP1 gene. It is characterized by the hypoplasia or absence of the cerebellar vermis. Signs and symptoms include rapid breathing (hyperpnea), sleep apnea, abnormal eye movements, mental retardation, and ataxia.