Infantile Neuroaxonal Dystrophy

National Cancer Institute

Source

National Cancer Institute. Infantile Neuroaxonal Dystrophy. NCI Thesaurus. Code C84927.

A rare autosomal recessive neurodegenerative disorder caused by mutations in the PLA2G6 gene. It is characterized by the development of swellings called spehroids along the axons of the central nervous system. Signs and symptoms appear early in life and include movement difficulties, muscle hypotonia and spasticity, and dementia.