Canavan Disease

National Cancer Institute

Source

National Cancer Institute. Canavan Disease. NCI Thesaurus. Code C84611.

A disorder that belongs in the group of leukodystrophies. It is caused by mutations in the ASPA gene which is responsible for the production of the enzyme aspartoacylase. It is characterized by spongy degeneration of the white matter of the brain. Signs and symptoms appear in infancy and include mental retardation, loss of motor skills, abnormal muscle tone, feeding difficulties and a very large head.