Canavan Disease

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Canavan Disease Information Page.

Canavan disease is a gene-linked neurological disorder in which the brain degenerates into spongy tissue riddled with microscopic fluid-filled spaces. Canavan disease has been classified as one of a group of genetic disorders known as the leukodystrophies. Recent research has indicated that the cells in the brain responsible for making myelin sheaths, known as oligodendrocytes, cannot properly complete this critical developmental task. Myelin sheaths are the fatty covering that act as insulators around nerve fibers in the brain, as well as providing nutritional support for nerve cells. In Canavan disease, many oligodendrocytes do not mature and instead die, leaving nerve cell projections known as axons vulnerable and unable to properly function. Canavan disease is caused by mutation in the gene for an enzyme called aspartoacylase, which acts to break down the concentrated brain chemical known as N-acetyl-aspartate.

Symptoms of Canavan disease usually appear in the first 3 to 6 months of life and progress rapidly. Symptoms include lack of motor development, feeding difficulties, abnormal muscle tone (weakness or stiffness), and an abnormally large, poorly controlled head. Paralysis, blindness, or hearing loss may also occur. Children are characteristically quiet and apathetic. Although Canavan disease may occur in any ethnic group, it is more frequent among Ashkenazi Jews from eastern Poland, Lithuania, and western Russia, and among Saudi Arabians. Canavan disease can be identified by a simple prenatal blood test that screens for the missing enzyme or for mutations in the gene that controls aspartoacylase. Both parents must be carriers of the defective gene in order to have an affected child. When both parents are found to carry the Canavan gene mutation, there is a one in four (25 percent) chance with each pregnancy that the child will be affected with Canavan disease.