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What is Canavan disease?

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Canavan disease (CD) is a relatively rare, but always fatal, inherited, degenerative brain disorder that primarily affects children of eastern and central European Jewish ([Ashkenazi](#)) descent, which includes about 90 percent of the Jews in America. It is estimated that one in 40 Ashkenazi Jews is a [carrier](#) of the Canavan gene.

The disorder was named for Myrtelle Canavan, the researcher who first described the disease in 1931.

Currently, there is no cure for the disease, which usually becomes apparent when the infant is three to nine months old. Symptoms vary, but generally include rapidly increasing head circumference, lack of head control, reduced visual responsiveness and abnormal muscle tone such as stiffness or floppiness.

Children
with
Canavan
disease
cannot

Symptoms

crawl,
walk, sit
or talk.
Over
time
they
may

- Rapidly increasing head circumference
- Lack of head control
- Reduced visual responsiveness
- Abnormal muscle tone

suffer seizures, become paralyzed, mentally retarded or blind and have trouble swallowing. Although hearing usually remains a functioning sense, deafness may also result. Most children do not live past age 10.

Canavan disease, like the more well-known Tay-Sachs disease, is one of a number of genetic disorders affecting Ashkenazi Jews at high frequency.

Information about some of these other Jewish genetic disorders (including Bloom syndrome; Familial Dysautonomia; Fanconi Anemia (Type C); Gaucher disease; Mucopolysaccharidosis IV; Niemann-Pick disease; Tay-Sachs disease) can be accessed through the websites of the National Foundation for Jewish Genetic Diseases, Inc. (www.nfjgd.org) and the National Tay-Sachs & Allied Diseases, Inc. (www.ntsad.org).

What several of these disorders have in common with Canavan disease is the lack of an essential enzyme that can lead to the abnormal accumulation of certain substances in the cells, causing cellular damage that results in the

degeneration of various functions.

Canavan disease is classified as one of the **leukodystrophies**—a group of inherited neurological disorders that affect the growth of the **myelin sheath**, the "white matter" of the brain that serves as an insulator to protect nerves.

Canavan disease is caused by a deficiency of the enzyme, **aspartoacylase** (ASPA), which leads to the buildup of **N-acetylaspartic acid** (NAA) in the brain.

The accumulated NAA causes a chemical imbalance that destroys the myelin, making the "white matter" spongy and resulting in Canavan disease's incapacitating symptoms.

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