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# Canavan disease

Canavan disease is a condition that affects how the body breaks down and uses aspartic acid.

## Causes

Canavan disease is passed down (inherited) through families. It is more common among the Ashkenazi Jewish population than in the general population.

The lack of the enzyme aspartoacylase leads to a buildup of material called N-acetylaspartic acid in the brain. This causes the white matter of the brain to break down.

There are two forms of the disease:

- Neonatal (infantile) -- This is the most common form. Symptoms are severe. Babies seem to be normal the first few months after birth. By 3 to 5 months, they have developmental problems, such as those mentioned below under the Symptoms section of this article.
- Juvenile -- This is a less common form. Symptoms are mild. Developmental problems are less severe than those of the neonatal form. In some cases, the symptoms are so mild that they go undiagnosed as Canavan disease.

## Symptoms

Symptoms often begin in the first year of life. Parents tend to notice it when their child is not reaching certain developmental milestones, including head control.

Symptoms include:

- Abnormal posture with flexed arms and straight legs
- Food material flows back into the nose
- Feeding problems
- Increasing head size
- Irritability
- Poor muscle tone, especially of the neck muscles
- A lack of head control when baby is pulled from a lying to a sitting position
- Poor visual tracking, or blindness

- Reflux (gastrointestinal) with vomiting
- Seizures
- Severe intellectual disability
- Swallowing difficulties

## Exams and Tests

A physical exam may show:

- Exaggerated reflexes
- Joint stiffness
- Loss of tissue in the optic nerve of the eye

Tests for this condition include:

- Blood chemistry
- Cerebrospinal fluid (CSF) chemistry
- Genetic testing for aspartoacylase gene mutations
- Head CT scan
- Head MRI scan
- Urine or blood chemistry for elevated aspartic acid
- DNA analysis

## Treatment

There is no specific treatment available. Supportive care is very important to ease the symptoms of the disease. Lithium and gene therapy are being studied.

## Support Groups

More information and support for people with Canavan condition and their families can be found at:

- National Organization for Rare Disorders -- rarediseases.org/rare-diseases/canavan-disease [https://rarediseases.org/rare-diseases/canavan-disease]
- National Tay-Sachs & Allied Diseases Association -- www.ntsad.org/index.php/the-diseases/canavan [https://www.ntsad.org/index.php/the-diseases/canavan]

## Outlook (Prognosis)

With Canavan disease, the central nervous system breaks down. People are likely to become disabled.

Those with the neonatal form often do not live beyond childhood. Some children may live into their teens. Those with the juvenile form often live a normal lifespan.

## Possible Complications

This disorder may cause severe disabilities such as:

- Blindness
- Inability to walk
- Intellectual disability

## When to Contact a Medical Professional

Contact your health care provider if your child has any symptoms of Canavan disease.

## Prevention

Genetic counseling is recommended for people who want to have children and have a family history of Canavan disease. Counseling should be considered if both parents are of Ashkenazi Jewish descent. For this group, DNA testing can almost always tell if the parents are carriers.

A diagnosis may be made before the baby is born (prenatal diagnosis) by testing the amniotic fluid, the fluid that surrounds the womb.

## Alternative Names

Spongy degeneration of the brain; Aspartoacylase deficiency; Canavan - van Bogaert disease

## References

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