What is Canavan disease?

Canavan disease is a type of leukodystrophy.

Leukodystrophies are a group of genetic conditions that mainly affect the white matter of the brain and spinal cord. The white matter is the wiring network of the brain that connects different brain regions and links the brain to the spinal cord and rest of the body.

Based on the onset of the condition and rate of progression, Canavan's disease is classified into:

1. **Neonatal/Infantile Canavan disease:** An early onset severe form that affects infants and causes deterioration of their development (regression).
2. **Juvenile Canavan disease:** This is a less common type of this condition that has milder symptoms.

What are symptoms associated with Canavan disease?

1) **Neonatal/infantile form:** Children initially develop normally but start showing symptoms at around 5 months of age with children not attaining age-appropriate milestones and losing the skills that they have acquired. Symptoms include:
   - Poor muscle tone.
   - Developmental delay.
   - Larger head size compared to other children of their age.
   - Feeding difficulties with or without difficulty in swallowing.
   - Stiffness of joints and muscles.

2) **Juvenile form:** Affected individuals have mildly delayed development of speech and motor skills starting in childhood. These delays may be so mild and nonspecific that they are never recognised as being caused by Canavan disease.

What causes Canavan disease?

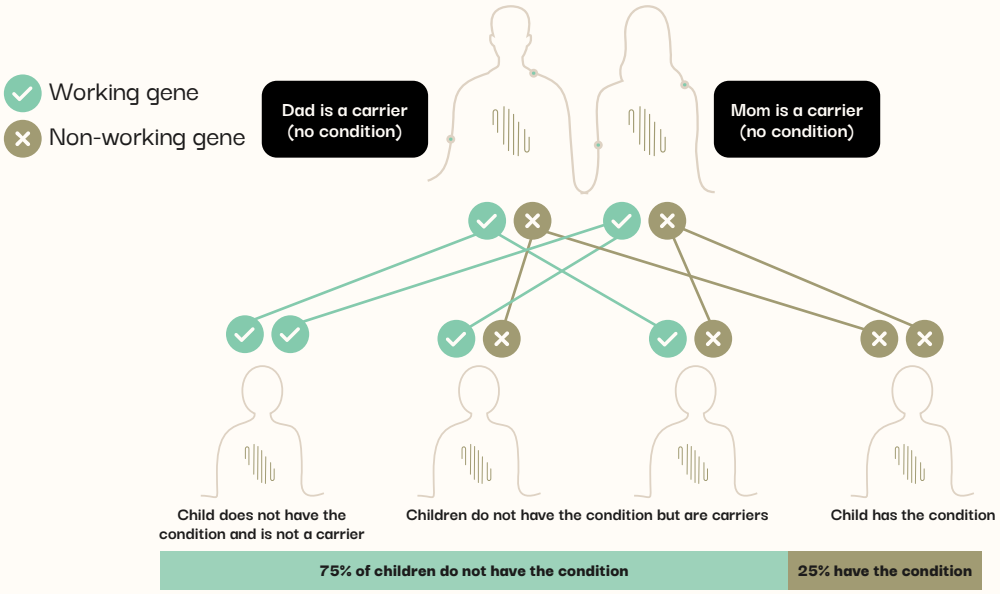
Canavan disease is caused by changes in the *ASPA* gene. This results in accumulation of N-acetyl aspartic acid (NAA) in brain tissue, which damages the white matter.

How is Canavan disease diagnosed?

- Clinical features: Typical clinical findings such as developmental delay, regression, poor muscle tone and gastrointestinal issues.
- Brain MRI: shows a characteristic pattern of white matter abnormalities.
- Elevated NAA in urine.
- Genetic testing confirming changes in the *ASPA* gene.

How is Canavan disease inherited?

Canavan disease in inherited in an autosomal recessive pattern. This is where two copies of the altered *ASPA* gene change causes Canavan disease. Each parent passes on one copy of the altered gene change.



Can Canavan disease be treated?

Treatment for early onset Canavan disease is supportive care directed towards assisting the families in alleviating symptoms. There is no specific treatment or cure. For individuals with mild/juvenile form of Canavan's disease, speech therapy or focused help with education might be required in some individuals. Researchers are looking at new ways to treat Canavan disease including gene therapies.

Support and resources:

- **Canavan Foundation** canavanfoundation.org/home
- **Canavan Research Foundation** canavan.org
- **Canavan Research Illinois** canavanresearch.org
- **Leukodystrophy Australia** leuko.org.au
- **Mission Massimo Foundation** missionmassimo.com
- **United Leukodystrophy Foundation** ulf.org/leukodystrophies
- **Hunter's Hope** huntershope.org/familycare/leukodystrophies

Research:

- **Australian Leukodystrophy Clinical and Research Program** leukonet.org.au
- **Clinical trials** clinicaltrials.gov/ct2/results?cond=canavan&term=&cntry=&state=&city=&dist=
- **Global Leukodystrophy Initiative** theglia.org

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