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COMMENTARY

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# Treatment of Canavan Disease and its Causes

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## Description

Canavan disease or Canavan-Van Bogaert-Bertrand disease is a rare and fatal autosomal recessive-generative disorder that causes progressive damage to nerve cells and loss of white matter in the brain. It is one of the most common degenerative brain diseases of infancy. It is caused by a deficiency of the aminoacylase 2 enzyme and is one of a group of genetic diseases known as leukodystrophy. It is characterized by the degeneration of myelin in the phospholipid layer insulating the axon of a neuron and is linked to a gene located on human chromosome 17.

Neonatal/childhood Canavan disease is the most common and most severe form of the disease. Affected children appear normal for the first few months of life, but between 3 and 5 months of age, developmental problems become apparent. These children typically do not develop motor skills such as rolling over, controlling head movement, and sitting without support. Other common features of the condition include weak muscle tone (hypotonia), unusually large head size (macrocephaly), and irritability. Feeding and swallowing difficulties, seizures and sleep disturbances may also develop.

The mild/juvenile form of Canavan's disease is less common. Affected individuals have slightly delayed speech and motor development since childhood. These delays can be so mild and nonspecific that they are never recognized as being caused by Canavan disease.

### Treatment

There is no known cure for Canavan disease, and there is no standard treatment procedure. Treatment is symptomatic and supportive. Physical therapy can help improve motor skills, and educational programs can help improve communication skills. Seizures are treated with antiepileptic drugs and a gastrostomy is used to maintain adequate food intake and hydration when there is difficulty swallowing. Experimental treatment also uses lithium citrate. When a person has Canavan disease, their levels of N-acetyl aspartate are chronically elevated. Lithium citrate has been shown in a genetic rat model of Canavan disease to be able to significantly reduce N-acetylaspartate levels. In human testing, the subject's condition reversed during a 2-week washout period after withdrawal of lithium.

The research revealed both reduced levels of N-acetylaspartate in the brain regions tested, as well as magnetic resonance imaging values that are characteristic of normal development and myelination. This evidence suggests that a larger controlled study of lithium may be warranted as supportive therapy for children with Canavan disease.

Experimental results of gene therapy published in 2002 used a healthy gene to take over the defective gene that causes Canavan disease. In human studies, the results of which were published in 2012, this method was shown to improve the patient's life without long-term adverse effects during a 5-year follow-up.

### Causes

Mutations in the ASPA gene cause Canavan disease. The ASPA gene provides the instructions for making an enzyme called aspartoacylase. This enzyme normally breaks down a compound called N-acetyl-L-aspartic acid (NAA), which is found mainly in neurons in the brain. The function of NAA is unclear. Researchers suspected that it played a role in the production of the myelin sheath, but recent studies suggest that NAA does not have this function. The enzyme may instead be involved in the transport of water molecules from neurons.

Mutations in the ASPA gene reduce the function of aspartoacylase, which prevents the normal breakdown of

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NAA. The mutations that cause the neonatal/childhood form of Canavan disease severely disrupt the enzyme's activity, allowing NAA to accumulate in the brain to high levels. Mutations that cause a mild/juvenile form of the disorder have milder effects on enzyme activity, resulting in less NAA accumulation.

Excess NAA in the brain is associated with signs and symptoms of Canavan disease. Studies suggest that if NAA is not properly broken down, the resulting chemical imbalance disrupts the formation of the myelin sheath as the nervous system develops. Accumulation of NAA also leads to the gradual destruction of existing myelin sheaths. Without this protective covering, nerves fail, disrupting normal brain development.