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CARRIER SCREEN

Canavan disease

What is Canavan disease?

Canavan disease is an inherited disease characterized by large head size, lack of head control and developmental delay. Signs and symptoms of Canavan disease are usually recognized by three to five months of age.[1] Individuals with Canavan disease have a deficiency in the aspartoacylase enzyme, leading to the accumulation of N-acetylaspartic acid in brain tissue.[2]

What are the symptoms of Canavan disease and what treatment is available?

Canavan disease most frequently occurs as a severe infantile disease with symptoms that include:[1]

- Progressive and severe developmental delay and intellectual disability
- Progressive hypotonia (low muscle tone) and later spasticity (becoming stiff)
- Macrocephaly (increased head size)
- Lack of head control
- Lack of developmental milestone achievements
- Loss of vision (due to optic atrophy)
- Seizures
- High levels of N-acetylaspartic acid in urine, blood and cerebrospinal fluid

Juvenile Canavan disease has been reported with a milder course that can include normal or mildly-delayed speech and motor development.[1]

There is no cure for Canavan disease. Individuals diagnosed with the infantile form have a shortened lifespan, typically into their teenage years. Treatment includes supportive care for symptoms.[1]

How is Canavan disease inherited?

Canavan disease is an autosomal recessive disease caused by mutations in the *ASPA* gene.[1] An individual who inherits one gene mutation in the *ASPA* gene is a carrier of Canavan disease and is not expected to have related health problems. An individual who inherits two mutations in the *ASPA* gene, one from each parent, is expected to be affected with Canavan disease.

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy. Therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Canavan disease?

Canavan disease can occur in individuals of any ethnic background, but is most common in individuals of Ashkenazi (Eastern European) Jewish ancestry. The carrier frequency of Canavan disease in the Ashkenazi Jewish population is estimated to be 1 in 55, with a calculated disease incidence of 1 in 12,100.[3]

Having a relative who is a carrier or is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier may also be influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

Canavan Research Foundation: http://www.canavan.org/

Canavan Foundation: http://www.canavanfoundation.org/about_canavan_disease

National Tay-Sachs & Allied Diseases (NTSAD): http://www.ntsad.org/

References

1. Matalon R *et al*. Canavan Disease. http://www.ncbi.nlm.nih.gov/books/NBK1234/. Accessed February 23, 2012.
2. Canavan disease. http://ghr.nlm.nig.gov/condition/canavan-disease. Accessed March 28, 2012.
3. Scott SA *et al*. Experience with Carrier Screening and Prenatal Diagnosis for 16 Ashkenazi Jewish Genetic Diseases. *Hum Mut*. 2010; 31(11): 1240-1250.