Canavan Disease (ASPA)

Canavan disease is a disease affecting the brain, caused by defects in the ASPA gene. The gene encodes for the enzyme aspartoacylase, whose function is to decompose excess N-acetyl-L-aspartic acid (NAA) in the brain. If the enzyme fails to function, excess NAA interferes with the development of the myelin sheath, the insulating covering around axons which functions to increase speed of neural transmission. The most common form of Canavan Disease, the neonatal or infantile form, causes a failure to develop normal motor skills. They suffer from macrocephaly, hypotonia, and often irritability. Seizures and difficulty swallowing may occur. Children rarely survive beyond their teens, and many die earlier. A milder form of the disease, the juvenile form, sometimes occurs. This is associated with slower than normal development of speech and motor skills, but does not normally lead to severe symptoms or a shortened lifespan.

Canavan disease is most common in those of Ashkenazi Jewish descent, where it is estimated to occur in 1 in 6,400 to 1 in 13,500 births. The incidence in the general population is much lower, but accurate estimates are not available. The disease is inherited as autosomal recessive, which typically requires both parents to be carriers of the faulty gene, most likely asymptomatically.

Sources

Matalon, R. & Michals-Matalon, K. (1999), “Canavan Disease,” in Pagon, R.A. et al., editors, GeneReviews [Internet]. See http://www.ncbi.nlm.nih.gov/books/NBK1234/

NIH, Genetics Home Reference: ASPA gene. See http://ghr.nlm.nih.gov/gene/ASPA

NIH, Genetics Home Reference: Canavan disease.
See http://ghr.nlm.nih.gov/condition/canavan-disease