Disability Awareness Begins With You:

Alexander Disease

What is Alexander disease?

Alexander disease is a rare disorder of the nervous system that usually begins before 2 years of age (infantile form). Onset can also occur, although less commonly, later in childhood (juvenile form) or adulthood. Alexander disease is considered one of the leukodystrophies, a group of disorders in which the primary abnormality is the inability to maintain the fatty covering (myelin) that insulates nerve fibers in the brain.

Signs and symptoms of the infantile form of Alexander disease usually include an enlarged brain and head (megalencephaly), stiffness in the arms and/or legs (spasticity), seizures, and mental and physical retardation. If disease onset occurs later in childhood, common problems include speech abnormalities, swallowing difficulties, and poor coordination (ataxia). Alexander disease is also characterized by abnormal deposits of proteins, called Rosenthal fibers, in specialized brain cells (astrocytes).

How common is Alexander disease?

The prevalence of Alexander disease is unknown. About 400 cases have been reported since the disorder was first described in 1949.

What genes are related to Alexander disease?

Mutations in the GFAP gene cause Alexander disease.

The GFAP gene produces a protein that is assembled inside the cell to make larger molecules (intermediate filaments) that are important for the normal activities of astrocytes. Mutations in the GFAP gene produce an altered protein, which is believed to block the assembly of intermediate filaments and other proteins. The blocked assembly of these proteins leads to their toxic accumulation in astrocytes, disrupting the normal functions of these cells. It is not yet clearly understood how impaired astrocytes contribute to the abnormal formation or maintenance of myelin.

How do people inherit Alexander disease?

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. Most cases result from new mutations in the gene and occur in people with no history of the disorder in their family. In some rare adult cases, the condition may be passed to children of the affected adult.

What other names do people use for Alexander disease?

- ALX
- AxD
- demyelinogenic leukodystrophy
- dysmyelinogenic leukodystrophy
- fibrinoid degeneration of astrocytes
- leukodystrophy with Rosenthal fibers

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