Familial encephalopathy with neuroserpin inclusion bodies

Description

Familial encephalopathy with neuroserpin inclusion bodies (FENIB) is a disorder that causes progressive dysfunction of the brain (encephalopathy). It is characterized by a loss of intellectual functioning (dementia) and seizures. At first, affected individuals may have difficulty sustaining attention and concentrating. They may experience repetitive thoughts, speech, or movements. As the condition progresses, their personality changes and judgment, insight, and memory become impaired. Affected people lose the ability to perform the activities of daily living, and most eventually require comprehensive care.

The signs and symptoms of FENIB vary in their severity and age of onset. In severe cases, the condition causes seizures and episodes of sudden, involuntary muscle jerking or twitching (myoclonus) in addition to dementia. These signs can appear as early as a person's teens. Less severe cases are characterized by a progressive decline in intellectual functioning beginning in a person's forties or fifties.

Frequency

This condition appears to be rare; only a few affected individuals have been reported worldwide.

Causes

FENIB results from mutations in the *SERPINI1* gene. This gene provides instructions for making a protein called neuroserpin, which is found in nerve cells (neurons). Neuroserpin plays a role in the development and function of the nervous system. This protein helps control the growth of neurons and their connections with one another, which suggests that it may be important for learning and memory.

Mutations in the *SERPINI1* gene result in the production of an abnormally shaped, unstable form of neuroserpin. Within neurons, defective neuroserpin proteins can attach to one another and form clumps called neuroserpin inclusion bodies or Collins bodies. These clumps disrupt the cells' normal functioning and ultimately lead to cell death. The gradual loss of neurons in certain parts of the brain causes progressive dementia. Researchers believe that a buildup of related, potentially toxic substances in neurons may also contribute to the signs and symptoms of this condition.

Learn more about the gene associated with Familial encephalopathy with neuroserpin inclusion bodies

- SERPINI1

Inheritance

FENIB is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In many cases, an affected person has a parent with the condition.

Other Names for This Condition

- Familial dementia with neuroserpin inclusion bodies
- FENIB

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Familial encephalopathy with neuroserpin inclusion bodies (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1858680/)

Genetic and Rare Diseases Information Center

- Familial encephalopathy with neuroserpin inclusion bodies (https://rarediseases.info.nih.gov/diseases/10037/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Familial encephalopathy with neuroserpin inclusion bodies%22)

Catalog of Genes and Diseases from OMIM

- ENCEPHALOPATHY, FAMILIAL, WITH NEUROSERPIN INCLUSION BODIES; FENIB (https://omim.org/entry/604218)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28familial+encephalopathy+%5BTIAB%5D+AND+neuroserpin+%5BTIAB%5D%29+OR+%28fenib%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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