Neuroacanthocytosis

Neuroacanthocytosis is a rare movement disorder. It has adult and childhood types. In adults, onset of symptoms usually begins between ages 20 and 50. In children, onset is often seen in adolescence. But it may occur earlier. Adult varieties can involve the heart and immune system.

CAUSES

It is typically an inherited autosomal recessive disorder. The disorder is due to:

- Degeneration of the basal ganglia. This is a part of the brain that helps control movement.
- Loss of neurons in the brain and spinal cord.

It is more common in males than in females. Some types have been caused by specific gene defects. Parkinsonism has been linked to the disorder in some patients.

SYMPTOMS

Symptoms may include:

- Muscle weakness and atrophy.
- Progressive cognitive loss.
- Involuntary twisting movements of the body (*chorea*).
- Acanthocytosis. This is spiked red blood cells. It is linked to inherited neurological disorders.
- Facial tics.
- Uncontrolled muscle movement.
- Unstable walking.
- Seizures.
- Biting of the tongue and lips.
- Changes in:
  - Personality.
  - Comprehension.
  - Judgment.

TREATMENT

Treatment is symptomatic and supportive. Antipsychotic drugs that block dopamine can provide temporary relief from tics and chorea. Drugs used to decrease anxiety can also decrease movement disorders. Those disorders are often made worse by stress. Injections of botulinum toxin can relax muscles and reduce unintentional movement. Other drug therapy may include anticonvulsants and antidepressants. Proper nutrition is required. A feeding tube may be needed for some patients as the disorder gets worse. Speech therapy and physical therapy may provide some relief to some patients.

This is a progressive disease. It is often fatal. That is generally the result of symptoms that lead to:

- Pneumonia.
- Cardiomyopathy.
- Eating problems.
- Other complications.

Life expectancy following onset of severe symptoms is typically 5-10 years. But life span may be near normal for patients with no neurologic or cardiac complications.

RESEARCH BEING DONE

The NINDS supports research on these disorders. It is aimed at:

- Increasing scientific understanding of the disorders.
- Finding ways to prevent and treat them.

The molecular changes responsible for some types of this disorder have recently been found. Researchers are examining the role of the basal ganglia. They hope to correlate the specific genetic abnormalities with the clinical features of the disease. Other research is aimed at identifying possible causes of sudden death related to heart muscle abnormality in patients with this disorder.

Research is continually being done to learn more about this problem.

Provided by: The National Institute of Neurological Disorders and Stroke

Document Released: 12/08/2003 Document Re-Released: 11/30/2009
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