Pelizaeus-Merzbacher Disease

Pelizaeus-Merzbacher disease is a rare, progressive, degenerative central nervous system disorder. With it, coordination, motor abilities, and intellectual function deteriorate. This disease is one of a group of genetic disorders called the leukodystrophies. These affect growth of the myelin sheath. That is the fatty covering--which acts as an insulator--on nerve fibers in the brain.

CAUSES

It is usually caused by a mutation of the gene that codes for a myelin protein called proteolipid protein or PLP. There are several forms of this disease. They include classic, connatal, transitional, adult variants. Except for the adult form of the disease, onset is usually in early infancy.

SYMPTOMS

- Early symptoms may include:
  - Slow growth.
  - Rapid, involuntary, rhythmic jerking of the eyes and the head This is called *nystagmus*.
  - Failure to develop normal control of head movement.
- In adult-onset cases, deteriorating speech may be an early sign.
- Other symptoms may include:
  - Tremor.
  - Various involuntary movements.
  - Grimacing.
  - Weakness.
  - Unsteady gait.
  - Shrinkage or shortening of a muscle. This is called *muscle contractures*.
- Over time, legs and arms may become spastic. Mental functions may decrease.
- Some patients may have convulsions. Skeletal deformation may result from abnormal muscular stress on bones.

TREATMENT

There is no cure for this disease. There is no standard course of treatment either. Treatment is symptomatic and supportive. It may include medication for seizures and movement disorders.

The prognosis for patients with this disease is poor. The course of the disorder is usually progressive deterioration until death.

RESEARCH BEING DONE

NINDS supports research on genetic disorders. This includes the leukodystrophies. The goals of this research are:

- To increase scientific understanding of these disorders.
- To find ways to prevent, treat, and, ultimately, cure them.

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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