Adrenoleukodystrophy

Adrenoleukodystrophy (ALD) is a rare, genetic disorder. It is characterized by the breakdown or loss of the myelin sheath surrounding nerve cells in the brain and progressive dysfunction of the adrenal gland. ALD is one of a group of genetic disorders called the leukodystrophies. These cause damage to the myelin sheath. It is a fatty covering - which acts as an insulator - on nerve fibers in the brain.

FORMS OF ALD

Classic childhood form. This is the most severe form and affects only boys. Its onset may occur between ages 4 and 10. Features of this form may include:

- Visual loss.
- Learning disabilities.
- *Seizures* (convulsions).
- Dysarthria (poorly articulated speech).
- Dysphagia (difficulty swallowing).
- Deafness.
- Disturbances of gait and coordination.
- Fatigue.
- Vomiting.
- Melanoderma (increased skin pigmentation).
- Progressive dementia.

The most common *symptoms* (problems) usually include:

- Behavioral changes such as abnormal withdrawal or aggression.
- Poor memory.
- Poor school performance.

Adult-onset form. This is the milder form. It typically begins between ages 21 and 35. Symptoms may include:

- Leg stiffness.
- Progressive spastic paraparesis (stiffness, weakness and/or paralysis) of the lower *extremities* (arms/hands and legs/feet).
- *Ataxia* (uncoordinated or unsteady movements) or (lack of muscle control).

Although adult-onset ALD progresses more slowly than the classic childhood form, it can also result in deterioration of brain function.

Another form of ALD is occasionally seen in women who are carriers of the disorder. Symptoms are mild and may include:

- Spastic paraparesis of the lower limbs.
- Ataxia.
- Hypertonia (excessive muscle tone).
- Mild peripheral neuropathy.
- Urinary problems.

Neonatal ALD. This affects both male and female newborns. Symptoms may include:

- Mental retardation.
- Facial abnormalities.
- Seizures.
- Hypotonia (low muscle tone).
- Heptomegaly (enlarged liver).
- Retinal degeneration.
- Adrenal dysfunction

This form usually progresses (worsens) rapidly.

TREATMENT

Bone marrow transplantation is effective early in the course of the childhood form. Physical and psychological therapy is important in all forms of ALD.

WHAT WILL HAPPEN

Prognosis (likely outcome) for patients with ALD is generally poor except after successful bone marrow transplantation. Death may occur within 1 to 10 years after the onset of symptoms due to disease progression.

RESEARCH BEING DONE

The NINDS supports research on genetic disorders such as ALD. The aim of this research is to find ways to prevent, treat, and cure these disorders.

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