Newborn Screening Provider ACT Sheet

Adrenoleukodystrophy, X-ALD

Adrenoleukodystrophy is a hereditary disease linked to the X chromosome. It is a result of fatty acid buildup caused by the relevant enzymes not functioning properly, which then causes damage to the myelin sheath of the nerves in the brain. If a diagnosis is suspected, a blood test of very long-chain fatty acids will detect elevated levels in 99% of males.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

Diagnostic Evaluation:

- Contact family to inform them of the newborn screen results. Point out additional tests are required to confirm the diagnosis
- Consult with a medical professional who is a specialist in genetic testing.
- X-ALD is suspected in four situations:
    - Boys with Attention deficit disorder (ADD)
    - Young men with progressive trouble walking or coordinating movements
    - All males with adrenal insufficiency (Addison disease) even in the absence of other symptoms
    - Adult women with progressive muscle weakness or wasting

Clinical Considerations:

Treatment depends on the signs and symptoms present in each person which may include:

- Corticosteroids in case of adrenal insufficiency which are used to normalize hormone levels
- Brain MRI's to determine extent of the disease
- Physical therapy
- Bone Marrow transplant

Additional Information:

X-ALD is inherited in an X-linked manner. Females have two X chromosomes and each male has an X and a Y chromosome. If the ABCD1 gene, has a disease-causing change, they will have X- ALD. Women who have disease-causing changes in one copy of the ABCD1 gene, are known as carriers of the disease. About 80% of carriers do not have signs or symptoms of X- ALD, because they have another working copy of ABCD1. If a male is diagnosed with X-ALD, it is likely that his mother is a carrier of the disease. If a woman is found to be a carrier, for each of her children there is a 50% chance that he or she will inherit the change of ABCD1.

For more information:

Genetic Home Reference (GHR)
Medline Plus
The National Institute of Neurological Disorders and Stroke
The National Organization for Rare Disorders