TEXAS Health and Human Services | Texas Department of State Health Services

Newborn Screening ACT Sheet

Elevated C26:0 Lysophosphatidylcholine

X-linked Adrenoleukodystrophy (X-ALD)

Differential Diagnosis

X-linked adrenoleukodystrophy (X-ALD), other peroxisomal disorders (including Zellweger spectrum disorders).

Condition Description

X-ALD is an X-linked genetic disorder caused by a defect in the adrenoleukodystrophy protein (ALDP) causing the accumulation of abnormally high levels of very long chain fatty acids in the body. This affects the nervous system white matter and the adrenal cortex. There are 3 variants of X-ALD: a childhood cerebral form that occurs primarily in males, adrenomyeloneuropathy (AMN), and Addison-only disease. Female carriers can develop AMN but typically onset is later and milder than in affected males.

Take the Following IMMEDIATE Actions

- **Contact family** to inform them of the newborn screening result and ascertain clinical status. No clinical signs are expected in newborns with confirmed X-ALD. The presence of symptoms (poor feeding, bony abnormalities, abnormal liver function testing, hypotonia, renal cysts) in a newborn may be suggestive of another peroxisomal disorder;
- **Consult** with pediatric *metabolic specialist* or *pediatric neurogeneticist*;
- **Evaluate the newborn**. If any sign (above) is present or infant is ill, transport to hospital for further evaluation and treatment in consultation with metabolic/genetic specialist;
- **Initial labs: Collect Very Long Chain Fatty Acids (CPT Code 82726).** *Draw specimen prior to feeding or 2-3 hours after feeding. The specialist may recommend additional testing;*
- Provide family with basic information about X-ALD disease, and signs and symptoms;
- Repeat newborn screen if second screen has not been done; and
- Report findings to newborn screening program.

Diagnostic Evaluation

Confirmatory very long chain fatty acid analysis. Patients with elevated values indicative of X-ALD or a peroxisomal disorder should have follow-up molecular genetic testing. Female carriers may also be identified.

Clinical Considerations

The childhood cerebral form of X-ALD manifests in males most commonly at around 4 to 10 years of age with attention deficit hyperactivity disorder, progressive cognitive and behavioral changes, adrenal impairment, and characteristic MRI abnormalities. X-ALD is caused by mutations in the ABCD1 gene and has an estimated incidence of approximately 1 in 17,000 live births. Adrenal steroid replacement is essential for treating adrenal insufficiency, however it doesn't prevent the development or the progression of neurological symptoms. Hematopoietic stem cell transplantation is the only proven successful treatment for the cerebral form of X-ALD but has to be performed in the early stages of the childhood cerebral form to be effective.

Additional Information

U.S. National Library of Medicine, Medline Plus – X-linked adrenoleukodystrophy
National Center for Biotechnology Information – Adrenoleukodystrophy
Baby's First Test – Adrenoleukodystrophy

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