Diagnosis, Monitoring & Care

How ALD is found, tracked, and treated — and how to build the right support around you.

DIAGNOSIS & NEWBORN SCREENING

ALD is diagnosed with genetic testing of the *ABCD1* gene and/or a blood test showing elevated very long-chain fatty acids (VLCFAs). Importantly, VLCFAs are falsely negative in 15% of females with ALD, thus genetic testing is the gold standard for diagnosis. Brain MRI is used to detect cerebral ALD before symptoms develop and to monitor for disease progression. Newborn screening identifies many babies at risk before symptoms appear. Although a positive newborn screen is not a diagnosis, it prompts confirmatory testing and early medical follow-up.

WHAT ONGOING MONITORING LOOKS LIKE

Because the course of ALD is unpredictable, boys are monitored closely throughout childhood. Brain MRI is typically performed every 6 months between ages 2 and 12, when cerebral ALD is most likely to develop, and annually thereafter. Adrenal function (cortisol and ACTH) is assessed regularly throughout childhood to detect adrenal insufficiency before symptoms develop. Monitoring continues in adulthood: men with ALD should have a brain MRI and adrenal testing every year, even without symptoms, because cerebral ALD and adrenal insufficiency can still develop. Women do not need routine brain MRI or adrenal testing unless they develop symptoms. Brain MRIs should be reviewed by an ALD specialist or neuroradiologist familiar with cerebral ALD. International recommendations for the monitoring of ALD are available[1], which outline currently recommended monitoring schedules to detect disease early.

TREATMENT & MANAGEMENT

Care is tailored to how ALD affects each person and may include:

Adrenal insufficiency: Daily hydrocortisone replacement, with higher ("stress") doses during illness, injury, or surgery.

Early cerebral ALD: Hematopoietic cell transplantation (HCT; also called a bone marrow or stem cell transplant) is most effective when performed at the earliest signs of disease, guided by brain MRI and the Loes score. Gene therapy may be an option for boys ages 4–17 with early cerebral ALD who do not have a suitable matched donor. Your transplant team can explain its benefits and risks, including an increased risk of blood cancer.

AMN and Symptomatic Women with ALD:
Physical therapy, rehabilitation, symptom management, and supportive care to maintain function and quality of life. Clinical trials may be an option; ask your care team or visit clinicaltrials.gov.

SIGNS THAT NEED PROMPT ATTENTION

- **Signs of adrenal crisis:** Severe fatigue, nausea or vomiting, dizziness, fainting, severe weakness, or salt craving require urgent medical evaluation.
- **New neurological symptoms:** New problems with walking, balance, coordination, weakness, or bladder or bowel control should be evaluated promptly.
- **In children:** Changes in behavior, school performance, vision, hearing, speech, or coordination may be early signs of cerebral ALD and should be reported immediately.

In an Emergency: An adrenal crisis is a life-threatening medical emergency. People with adrenal insufficiency should carry emergency hydrocortisone and wear a medical alert bracelet or other medical identification. Develop an emergency plan with your endocrinologist for illness, injury, surgery, or other significant physical stress.

General educational information, not medical advice — always consult a qualified ALD specialist.

BUILDING YOUR CARE TEAM

Endocrinologist: Monitors adrenal function and manages hormone replacement therapy and stress dosing.

Neurologist: Monitors the brain and spinal cord, reviews brain MRIs, and manages neurological symptoms.

Transplant Physician: Evaluates and treats people with cerebral ALD who may benefit from stem cell transplantation or gene therapy and, in some centers, provides long-term follow-up.

Genetic Counselor: Explains inheritance, coordinates family testing, and discusses reproductive options.

Urologist: Manages bladder issues and sexual dysfunction, which are common in adults with ALD.

Gastroenterologist: Manages bowel symptoms, which are common in adults with ALD.

Reference:

1. Engelen, M., et al. (2022). International Recommendations for the Diagnosis and Management of Patients With Adrenoleukodystrophy: A Consensus-Based Approach. Neurology, 99(21), 940–951. https://doi.org/10.1212/WNL.0000000000201374. PMID: 36175155.

Physical Therapist: Supports mobility, strength, balance, and independence.

Occupational Therapist: Helps maintain independence with daily activities and recommends adaptive equipment when needed.

Mental Health Professional: Provides emotional support and helps individuals and families cope with the challenges of ALD.

Social Worker: Connects families with community resources, insurance and disability services, financial assistance, and support programs.

QUESTIONS TO ASK YOUR CARE TEAM

- Which form(s) am I (or my child) at risk for, and how will you monitor them?
- How often should we have a brain MRI and adrenal (cortisol) testing?
- Should family members be tested? Is genetic counseling available?
- Where is the nearest ALD Center of Excellence?

QUESTIONS OR COMMENTS?
Visit ALDConnect.org or email us at info@aldconnect.org

ALDConnect