Provider action sheet for adrenoleukodystrophy (ALD)

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for adrenoleukodystrophy (ALD) or a related condition.

What steps do you need to take?

1. **Consult** with a California Children's Services Special Care Center (SCC) metabolic specialist. They will ask for your assessment of the infant's current health status and any family history of the disorder. The SCC specialist will arrange for a visit for further testing and multifaceted monitoring as needed.
2. **Contact** the family to explain the positive newborn screening result. For information on how best to communicate, consult the **Health Resources & Services Administration (HRSA) Newborn Communication Guide** (https://bit.ly/HRSAGuide or search online for "HRSA" "heritable" "communication")
3. **Review** the accompanying, "Family action sheet for adrenoleukodystrophy" with the family and ensure they understand.
4. **Inform** the family that this result requires confirmatory testing and evaluation. Emphasize that you, with ASC staff and the SCC specialists, will guide them through the next steps of confirmatory testing and follow-up services.
5. **Advise** parents to (1) follow the plan for confirmatory testing, and (2) keep their infant's appointments with the SCC specialists. The family should receive services from a team of specialists, including genetic counseling services.

Clinical information

Newborn screening identifies individuals who may have ALD or another peroxisomal disorder. There are multiple forms of ALD, including cerebral ALD (CALD), 'Addison disease only', and Adrenomyeloneuropathy (AMN).

X-linked ALD is an inherited peroxisomal disorder in which accumulation of very long chain fatty acids (VLCFA) in the brain, spinal cord, and adrenal glands leads to demyelination in the brain and spinal cord and impaired adrenal corticoid function in the adrenal cortex.

ALD is characterized by progressive physical, behavioral, cognitive, and neurologic deficits. While ALD primarily affect males, females could have symptoms, usually later in life. Close monitoring by metabolic and endocrine specialists' aids in early intervention and treatment to stop or slow disease progression.

Early and ongoing treatments may include stem cell transplantation, gene therapy, and corticosteroid treatment.

Please visit these sites. Search for the site name and "ALD" if needed.

- **GeneReviews** (https://www.ncbi.nlm.nih.gov/books/NBK1315/)
- **HRSA** (https://newbornscreening.hrsa.gov/conditions x-linked-adrenoleukodystrophy).

Questions?

For follow-up questions, please call your NBS Program Area Service Center (ASC).

For program questions, please email NBS Program staff at NBS@cdph.ca.gov or visit the NBS Program website (www.cdph.ca.gov/NBS).

California Department of Public Health
Newborn Screening Program (www.cdph.ca.gov/NBS)