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FAQ:
frequently asked questions

“ Experience shows that the community is most successful in getting adrenoleukodystrophy added to newborn screening labs when the champion behind it is someone who is personally impacted by this disease. That story and experience is very powerful. ”

-BRAD ZAKES

ADVOCATE IN WASHINGTON





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What is ALD?

Adrenoleukodystrophy (ALD) is a rare, X-linked genetic disease that occurs in about 1:21,000 males and 1:17,000 newborns in the total population. Caused by an underlying genetic mutation in the ABCD1 gene, ALD affects the body's ability to create the protein that helps break down very long-chain fatty acids (VLCFAs). A buildup of VLCFAs can cause adrenal problems and potentially progress to causing brain damage (ALD that progresses to the brain is referred to as cerebral ALD).

Who is affected by ALD?

ALD occurs in about 1:21,000 males and 1:17,000 newborns in the total population. As an X-linked disease, ALD affects males more severely; however, development of myelopathic symptoms in women with ABCD1 mutations is common and age dependent, with most developing clinical manifestations of the disease by the age of 60 years.

What is NBS?

Newborn screening (NBS) is testing that occurs after a baby is born, at no cost to a family, to detect for serious, but treatable, medical conditions. What conditions are screened for varies across states in the United States and from country to country around the world.

Why is NBS important?

Early detection and diagnosis allow for early treatment, which can prevent or reduce the symptoms of conditions such as ALD.

Where can I find more information on what states and countries have active NBS programs for ALD?

For more information on where ALD is screened in the United States, visit:

- [**Baby's First Test**](#)
- [**Every Life Foundation**](#)
- [**ALD Alliance**](#)

For more information on where ALD is screened for in Europe and/or the United Kingdom, visit your country's leukodystrophy association or the Ministry of Health's website.

What is the RUSP?

The RUSP, or Recommended Uniform Screening Panel, is a list of disorders in the United States that the Secretary of the Department of Health and Human Services (HHS) recommends for states to screen as part of their state universal NBS programs. To be considered for inclusion in the RUSP, a disease is required to meet several criteria, including:

- *The disease must be serious*
- *The disease must have a reliable test*
- *A treatment or therapy must exist*

Each state individually decides which conditions to screen for through their individual NBS programs. ALD was added to the RUSP in February 2016.



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importance of NBS



NBS and ALD



what is advocacy?



NBS today



NBS panels: US, UK, & EU



getting involved



resources



FAQ



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Why aren't all conditions on the RUSP automatically tested at the state level in the United States?

Newborn screening programs are a public health initiative managed at the individual state level where the ultimate decision on whether or not to test for any given condition is left to each state. Although states often rely on the recommendations provided through the Recommended Uniform Screening Panel (RUSP), not all states align with the federal panel.

Why isn't ALD screened for in more countries?

As a relatively rare condition, adrenoleukodystrophy (ALD) is not widely known around the world. More advocates, like you, are needed to drive awareness and bring education to ministries of health, healthcare professionals, and lawmakers around the world to ensure early diagnosis for all individuals born with ALD.

You can learn more about ALD and start to identify patient advocacy organizations in the resources section of this tool kit.

CONCLUSION
Lending your voice to advocacy is one of most powerful things you can do for a cause. Your personal story can help stakeholders understand the impact that their decision-making can have for families living with ALD.



We want to hear from you. Contact patientadvocacy@bluebirdbio.com to share your advocacy story and suggestions for improving this resource to better meet advocates needs.



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