A close-up photograph of a baby's face and hands. The baby's hands are clasped together near their mouth, and their face is partially visible in the upper right corner. The background is a soft, textured surface, possibly a blanket. A large, bright blue brushstroke graphic is on the left side of the image, containing the text.

newborn screening for adrenoleukodystrophy:

AN ADVOCACY TOOL KIT





special thanks

Thanks to the undertakings of advocates and policymakers around the world, newborn screening (NBS) is an active and evolving landscape. As information progresses and efforts continue to enhance the reach of current NBS programs, please note that the content included in this tool kit is dated, and is up to date as of September 2020.

Throughout this tool kit, you will notice that specific reference is made to select countries. These countries were chosen as a representative reflection of where NBS efforts, policies, and processes are today, in 2020, and where pilot programs are potentially more robust.

We would like to thank the following advocates for their contributions to the NBS tool kit and for their dedication for supporting the adrenoleukodystrophy (ALD) community:





about the tool kit

Intro

This resource has been created in collaboration with patient advocacy organizations and advocacy leaders. Its purpose is to raise awareness of:

- **Adrenoleukodystrophy (ALD)**—pronounced uh-Dree-noh-loo-koh-DIS-truh-fee
- The **lifesaving opportunity** newborn screening (NBS) can provide
- The critical **importance of early detection** and monitoring for disease progression

Through this tool kit, we hope to enable advocates around the world to better understand NBS and to empower them in their ongoing efforts to promote screening for ALD. We are thankful to all of the many individuals and families who raise their voices around the world for NBS of ALD. Your voices matter.



Please note that this is an interactive PDF. You can click the numbers at the right of the page to jump from section to section. There is also a series of dots at the bottom of the pages which indicates your position within that section. The bird house icon in the top right corner of each page will take you back to this page. The chapter numbers on the right navigation bar will take you to the beginning of each chapter. Additionally you may notice that some terms and phrases have been bolded to highlight their potential importance.

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1

the importance of NBS for ALD:

a call to action from like-minded advocates around the world



“ **Never doubt that a small group of thoughtful, committed citizens can change the world; indeed, it's the only thing that ever has.** ”

- MARGARET MEAD

CULTURAL ANTHROPOLOGIST

1

the importance of NBS for ALD:

a call to action from like-minded advocates around the world

Newborn screening (NBS) for adrenoleukodystrophy (ALD) means possibly saving children's lives.

It is critical, primarily for boys, who are most severely affected by the condition, but also to a lesser extent for girls, for extended-family screening and future planning. As a rare, X-linked metabolic disease, ALD can lead to adrenal problems such as adrenal insufficiency and potentially more serious complications if not detected early and managed. Early diagnosis through NBS enables the parents of young boys to know, from a very early point in the child's life, that their child must be monitored for progression to a life-threatening form of the disease—cerebral ALD. **By finding this information out while a child is still a baby, doctors can help each family plan for the future.**

They can also realize that **there is hope**. NBS for ALD provides access to a 'window of opportunity' and allows for the timely commencement of available therapies. Early detection of cerebral ALD may allow doctors to treat the disease in time to save lives.

The following quotes are from advocates and parents who are advocating for the inclusion of ALD as part of NBS panels. **Their voices explain firsthand how important it is to screen newborns for ALD.**

“There are a number of ways you can use your voice to support the ALD community... We need to drive the message home that this doesn't just happen to one boy. This disease can be stopped, and the cost of screening is minimal.”

– ELISA SEEGER / advocate in New York

“To advocate for NBS, you don't need to start an organization or a foundation, you can be an individual or a family who is just passionate about this cause... As horrific and difficult as diagnosis may be for a family, just knowing gives them the benefit of time, to be able to monitor the disease and be in a position where they can take necessary medical intervention at the appropriate time. That in my mind is the key. [NBS] will save lives. I think it already is.”

– BRAD ZAKES / advocate in Washington

“I advocate by setting the goal to have NBS implemented and then ensuring that happens by establishing and changing systems...the health system, political system, and social system. I talk to geneticists. I talk to members of Parliament. I am always making a case for inclusion of NBS. I do anything and everything that I can at every level to raise awareness.”

– BOB WYBORN / advocate in Australia



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what is ALD and what is NBS? understanding the condition of adrenoleukodystrophy

“**Newborn screening is a very efficient program. It provides a screening to families who would otherwise have no idea that they needed to be aware of these diseases.**”

-ANNA GRANTHAM
ADVOCATE IN ALABAMA



2 what is ALD and what is NBS?

understanding the condition of adrenoleukodystrophy

What is ALD?

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

Everyone's journey with ALD is unique. Some individuals may only experience one manifestation of ALD whereas, for others, these manifestations may overlap during their lifetime.

There are a few main ways ALD can manifest:

- **Asymptomatic:** ALD without signs or symptoms
- **Adrenal insufficiency:** ALD that results in adrenal symptoms such as fatigue, loss of appetite, hyperpigmentation (skin darkening), and belly pain. Most boys with ALD will eventually develop adrenal insufficiency. Although adrenal insufficiency is manageable it can become life-threatening if it is not detected early
- **Adrenomyeloneuropathy (AMN):** ALD that commonly starts in adulthood and affects the spinal cord and nerves
- **Cerebral ALD:** ALD that progresses to affect the brain. The condition can progress very quickly, so early diagnosis is essential.

Cerebral ALD is a severe form of ALD that occurs in about 40% of boys with ALD. In cerebral ALD, the layer of myelin that protects nerves in the brain is broken down. If left undiagnosed or untreated, this can have severe effects. These effects can potentially include significant disabilities, such as an inability to speak or respond, blindness, or even death.



There are a few ways a child may be diagnosed with ALD:

- Adrenal symptoms
- Newborn screening (NBS)
- Family history
- Neurologic symptoms

For those who are not diagnosed through NBS, an endocrinologist or geneticist/genetic counselor can confirm an ALD diagnosis by measuring VLCFA levels in blood plasma and testing for a mutation in the *ABCD1* gene. ALD can also occur as a result of a spontaneous mutation. You may hear this form of ALD described as *de novo*.

Progression of cerebral ALD can be effectively stopped if the disease is detected early. **The only currently available treatment option for cerebral ALD is allogeneic-hematopoietic stem cell transplantation (HSCT)**, which optimally occurs at a very early presymptomatic, sometimes early symptomatic, stage of the disease. HSCT is most effective during a narrow therapeutic window, which is often missed due to late diagnosis or misdiagnosis. NBS can provide the earliest opportunity for detection. NBS for ALD provides access to a "window of opportunity" and allows for a timely commencement of treatment.



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What is NBS?

Newborn screening (NBS) is a public health program that screens newborn infants for certain disorders and conditions that may not be immediately apparent at birth. Although NBS identifies the possibility of these disorders, additional lab tests may be needed to confirm diagnosis. For the diseases that are included in NBS panels, early detection can result in manageable and lifesaving outcomes.

Variability in NBS programs is due to a number of factors, including proper funding, available laboratory equipment and resources, and prioritization of conditions included in the screening panel.

NBS starts with a simple blood test

Within a short span of time (roughly 2 to 5 days) **after a baby is born, a physician, nurse, or midwife pricks the newborn's heel (in a process called a heel stick or heel prick).** Drops of blood are then collected on a card partially made of filter paper (sometimes called a Guthrie card), which preserves the blood for testing. The card also contains a space for information about the baby and the baby's parents' contact information, which is usually filled out before the heel stick. Once the blood spots are dry, the card is ready to be sent to the identified laboratory for analysis. Many labs may keep and store these samples for future need and use. **This is critical, as many of the conditions screened for by NBS need to be diagnosed as quickly as possible.**



A newborn baby receives a heel stick for NBS testing.

Laboratories will analyze the provided samples through a variety of tests including tandem mass spectrometry (MS/MS). MS/MS is the key method for detection of inherited metabolic diseases, a system able to screen for a multitude of diseases very rapidly. With a single dried filter paper blood spot, MS/MS can identify more than 30 inherited metabolic disorders in around 2 to 3 minutes. Thereafter the lab will take time to review and confirm the outcome of the test before communicating the results.



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How does the NBS test for ALD work?

Screening for adrenoleukodystrophy (ALD) involves testing to determine whether there are elevated very long-chain fatty acid (VLCFA) levels. If the results of newborn screening (NBS) testing for ALD are positive, follow-up testing for confirmation is a necessary step before a diagnosis can be made. **Follow-up testing can include genetic testing that looks for alterations in the *ABCD1* gene**, as these mutations are the ultimate cause of ALD.

Both screening for elevated VLCFA levels and genetic testing for the *ABCD1* gene are very accurate in males. **However, neither test can predict what kind of ALD a child will develop.** In females, detection rates for ALD are 80%–85% with VLCFA screening and 100% with genetic testing.

When a positive result occurs, the lab notifies the family's physician and the physician contacts the family. The information provided at the time of diagnosis differs from state to state and country to country. Several patient advocacy organizations have developed helpful informational materials for families whose son or daughter is diagnosed with ALD through newborn screening. Learn more at ALDNewbornScreening.org.

“NBS is a very efficient program. It provides a screening to families who would otherwise have no idea that they needed to be aware of these diseases. This is very common with leukodystrophies because they are so rare. For leukodystrophies, early detection through NBS is really the hope for these families. It is the only way we can ensure these children have a chance at a healthier life.”

– ANNA GRANTHAM / advocate in Alabama

What happens after screening?

The NBS test used for ALD **first detects elevated VLCFA levels in a baby's blood at birth.** If VLCFA levels are high, the baby will have to undergo additional testing to confirm a diagnosis of ALD.

Once diagnosed, boys with ALD can undergo possible treatment for any adrenal symptoms they may experience. Most urgently, as there is currently no way to predict which boys with ALD will develop cerebral ALD, NBS allows boys to be identified and monitored before the onset of symptoms of cerebral ALD.

Magnetic resonance imaging (MRI), monitoring for the brain changes that indicate cerebral ALD, can lead to improved outcomes, such as life-saving treatment through timely hematopoietic stem cell transplant.

Monitoring using MRI

ALD progresses differently in every child. And although about 40% of boys will develop cerebral ALD, medical guidelines suggest that all boys with ALD should be monitored for progression by having regularly scheduled MRI. MRI provides the earliest opportunity to detect cerebral ALD before any symptoms appear. Early detection of cerebral ALD can have lifesaving results.



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what is advocacy?

making a real difference in people's lives

“ There are a number of ways you can use your voice to support the ALD community... We need to drive the message home that this doesn't just happen to one boy. This disease can be stopped, and the cost of screening is minimal. ”

-JANIS SHERWOOD
ADVOCATE IN CALIFORNIA



3 what is advocacy?

making a real difference in people's lives

The World Health Organization (WHO) defines advocacy as the process of influencing people to create change.

Advocacy may be broadly defined as a strategic series of actions that accomplish the following goals and is a fundamental concept for affecting governmental actions:

- **Speaking in favor of a particular cause**
- **Influencing and arguing for a change in public policy and public opinion**
- **Prompting a transformation**

Regardless how or where they start, all types of advocacy can be broken down into three different concepts: mission, action, and change. Whether you have an entire organization behind you or you are simply an individual, the purpose behind advocacy is to have a mission, take action, and engender change.

1 MISSION

Advocacy is an active process that starts with a common mission. Advocacy in all forms seeks to ensure that people in a society are able to raise their voices, defend and safeguard their rights, and have their views genuinely considered when decisions are being made about their lives.



Example: Your mission may be to see newborn screening for adrenoleukodystrophy implemented in your state or country.

2 ACTION

Taking action for advocacy involves taking part in a broad range of activities that include, but are not limited to, raising awareness through public education, campaign tactics, experience-based events, social media, researching, and lobbying. In a time when the network of digital platforms continues to expand, there are more ways than ever before to get involved and make your voice heard. Many families turn to social media first to begin their advocacy efforts.



PUBLIC SPEAKING

Language is one of the most powerful tools that we have, allowing us to create new connections, channel empathy, and influence the actions and decisions of political leaders who may not be familiar with how their decisions affect their constituents' lives. Advocacy groups and organizations often enhance their voice through public speaking and by generating an online presence. By doing so, they are able to convey their message and, ultimately, create a positive impact.

Public speaking may sound daunting, but there are tools and tips to help you along the way. Remember, practice makes perfect.



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CAMPAIGNS/EVENTS

Spreading a message through the use of campaign tactics, such as organizing a meeting in your neighborhood, is another way to generate traction and create a movement. Local grassroots events are a great way for advocacy groups to network and reach individuals on a larger scale to help make a difference.



LOBBYING

Lobbying is a method of advocacy that occurs when an individual or a private interest group works to influence legislation at the local, regional, or national level.

Rules and regulations around lobbying requirements and registrations may differ from state to state and country to country.



DEVELOP CHAMPIONS WITHIN POLICY MAKER CIRCLES

Advocacy is usually first approached by generating public awareness about a particular cause or issue. Once public awareness regarding the issue has gained traction, policy makers may then evaluate and potentially decide to champion the need to take action.



One way to raise public awareness about a particular cause is by leveraging social media platforms. See [Take Your Voice on Social](#)

3 CHANGE

Change doesn't happen overnight. It takes diligence, commitment, and a relentless passion to get a message out into the world, connect others through a single vision, and keep them engaged long enough to drive transformation. It may not be easy, but with a goal in mind and a plan in place, advocacy can have an impact on the lives of many and, ultimately, be the force for change. It's only when action is taken for a particular cause that its end goal becomes attainable.



Change can seem like a slow process, but our collective efforts are worth it. Since the 1960s, patient advocacy groups in the United States have been instrumental in efforts to mandate state testing of newborns. However, once the condition is included, all newborns who undergo newborn screening will be screened for that condition locally. This is a good indication for advocates around the world that change is really possible at the local level.



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Advocacy for NBS

Advocacy for newborn screening (NBS) has been a global concern and a continuous law-changing effort worldwide. Although some progress has been made, **NBS programs around the world still require improvements, and most still do not include adrenoleukodystrophy (ALD).** As countries decide which diseases to add to their programs, advocates around the world still play an incredibly important role in raising awareness about the need for expanded NBS.

“ I am someone who 10 years ago found out my son had ALD and was waiting for the other shoe to drop. I should buy a lottery ticket every day; I'm this lucky that we've had 10 years of good health and knowing and watching science progress... There's not one friend or colleague I have that has lost a child or family member to ALD who did not wish they knew one day sooner. It's frustrating that NBS is not available everywhere. It's always worth knowing.”

– KATHLEEN O'SULLIVAN FORTIN /
advocate in Massachusetts

Who advocates for NBS and how does it become actionable?

Adding a disorder to the NBS panel is done through a collaborative process that may involve relevant stakeholders and influencers such as genetic specialists, public institutions, health agencies, NBS laboratories, healthcare providers, patients' associations, and policy makers. Here is where your story and your voice can make an impact. As it relates to NBS advocacy, here are some stakeholders that you may want to reach, engage, and influence to make your case for NBS:

• General Stakeholders

- Families with ALD in your state or country
- Advocates for NBS for ALD and other rare diseases

• Influencers

- Media
- Patient/community advocates
- Healthcare professionals (public health officials, researchers, health agencies, etc)

• Specific NBS decision-makers

- Primary audience: Legislators/regulators, policy makers, Commissioner of Health
- Secondary audience: Medical professionals, laboratories



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where is NBS today?
the current status quo of newborn screening

“ When I discovered there was a newborn screening test available for adrenoleukodystrophy and not one state was utilizing it, I decided this is where I needed to put all of my energy. ”

- ELISA SEEGER
ADVOCATE IN NEW YORK

4 where is NBS today?

the current status quo of newborn screening

ALD and NBS

Currently, **Washington, DC and several states in the US include testing for adrenoleukodystrophy (ALD) as part of their newborn screening (NBS) panel**, whereas several other states are mobilizing efforts.

To find out whether ALD is part of NBS panels where you live, visit resources such as:

- [Baby's First Test](#)
- [ALD Alliance](#)
- [ALD Connect](#)
- [United Leukodystrophy Foundation](#)
- [EveryLife Foundation](#)



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Outside of the United States, only the Netherlands has approved the addition of ALD NBS (for boys) through a pilot launched in 2019. Although progress continues, there is still a lot of work to be done to ensure that newborns are screened for ALD.



Now we are at a point where [several] states are actively testing for ALD—Texas just started and that means 50% of all babies born in America are now tested for ALD—which is AMAZING!”

– KATHLEEN O’SULLIVAN FORTIN /
advocate in Massachusetts



What advocacy can do: the example of Aidan’s Law

Through the efforts of Aidan’s Law, an advocacy campaign dedicated to making ALD a part of every NBS panel in the United States, New York was the first state to approve the addition of ALD to its screening program on December 30, 2013. This addition was a direct result of the work of the ALD Alliance in New York and the advocacy efforts of families across the United States.



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4 where is NBS today?

the current status quo of newborn screening

As of today, newborn screening (NBS) programs around the world vary substantially, even within developed countries. Anywhere from **1 to approximately 50 conditions are included in NBS panels**. The **Recommended Uniform Screening Panel (RUSP)** in the United States is the most robust and comprehensive list of conditions recommended for NBS, currently suggesting screening for 35 core disorders and 26 secondary disorders; this is effective as of March 2020.

“ I advocate for myself, for my children, for my brothers, for my mother. We want to change the landscape of ALD, and we feel we all have unique gifts to give and share. Everyone can be an advocate and have a voice.”

- FRANI BROUSSARD / advocate in Texas

The reason why NBS panels vary greatly from country to country and from state to state are based on many factors, including **politics, culture, sociology, ethics, and certainly economics**. For example, as of 2018, the United Kingdom only screens for 9 rare, but serious, conditions, whereas the Italian NBS panel currently includes ~40 conditions. The differences between countries are substantial, causing advocates from several organizations to work with their governments to add more conditions to their NBS program.



The RUSP may be used to influence governing bodies in other regions and countries outside of the United States as proof of the necessity to include a condition on their NBS panels. The US Health Resources and Services Administration website provides information on the current conditions listed on the RUSP. It also provides information on the history of conditions that have been recommended or not recommended as additions to the RUSP. This information includes the data and support used to make cases for conditions to be added to the RUSP, which can be incredibly helpful for advocates who are looking to build a case for a condition to be added to the NBS program in their area.



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Adding a condition to any given panel is contingent on several factors, which may include:

- **Evidence** that supports the benefit of screening
- Availability of **effective treatments** for the condition
- **Cost implications** of launching a newborn screening (NBS) program (including cost-effectiveness)
- **Cultural understanding and acceptance**

Some of these factors, namely benefit of screening, availability of effective treatments, and cost implications analysis can be managed with data and information to support inclusion of a new condition on an NBS panel, whereas the cultural aspects could be more challenging. Through hard work and persistence, advocates can continue to overcome such challenges!

As an example, laboratories in a given country may not have the resources to perform a test for a given condition if they have not already been fully equipped to perform NBS services. The resource needs are often closely related with the cost of being able to employ the relevant personnel and/or to afford the specific lab equipment, such as the tandem mass spectrometer, which is the machine needed to run the tandem mass spectrometry.

As an advocate, you can play a role in helping decision-makers understand the cost-effectiveness of NBS for adrenoleukodystrophy (ALD). If evidence supports the fact that NBS for a certain condition can reduce or eliminate the costs of some healthcare expenditures, NBS for that disease will be deemed cost effective. If NBS for a certain condition should be deemed cost effective, public payers and governments will be more willing to add such a condition to the NBS panel and fund the programs to initiate the screening.

As it relates to ALD NBS, researchers have already conducted studies examining and analyzing its cost-effectiveness ratio. Typically, these studies involve the development of an economic model, eg, a decision tree model based in Microsoft Excel that considers all of the relevant benefits/disadvantages and costs associated with an NBS program.

The main components of a model **evaluating the cost effectiveness of NBS for ALD** should include the following data:

- 1 the epidemiology of ALD
- 2 the sensitivity and specificity of the screening test for ALD
- 3 the assumed rates of survival for ALD
- 4 the costs associated with screening and treatment of ALD



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Key issues that drive the results for cost-effectiveness analysis include:

- 1 the quality-of-life and behavioral impacts of receiving an **early diagnosis for adrenoleukodystrophy (ALD)** that may not lead to cerebral ALD
- 2 the quality-of-life and behavioral impacts of receiving an **early diagnosis for another peroxisomal disorder**, especially if no effective treatments are currently available for that disease
- 3 the potential **risk of false positives** (ie, an individual or their family is informed of having a disease when in fact they do not) **and false negatives** (ie, a family is informed that their infant does not have the disease when in fact he does)

Data sources used for the cost-effectiveness models typically include published literature and expert clinical judgment. Even if some analyses relating to the cost effectiveness of ALD newborn screening (NBS) already exist, countries may want to obtain results based on their own national and state-specific data. This doesn't mean that they won't be willing to accept or at least review already existing research.

The cost-effectiveness analysis of NBS for ALD in the UK did in fact conclude that screening for ALD would be a cost-effective use of UK healthcare resources, especially as the NBS program is projected to reduce lifetime costs and improve outcome for boys progressing to cerebral ALD. The favorable economic results were driven by estimated reductions in the social care and education costs. It is hoped that additional analyses will be published in the future as decision-makers in other countries assess NBS for ALD.



There's no need to start from scratch in your advocacy for NBS. Learn about the history of NBS in your area. Find out how other advocacy organizations have gotten conditions added to the NBS panel over time. The experience of other successful organizations can help to provide you with a road map for your **advocacy efforts**.



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how are diseases added to NBS panels in the United States, in the United Kingdom, and in select EU countries?

“Advocates should always be proactive, anticipate someone’s questions, and leave the door open for someone to get back to you when they’re ready to learn more.”

—BOB WYBORN
ADVOCATE IN AUSTRALIA



5

how are diseases added to NBS panels in the United States, in the United Kingdom, and in select EU countries?

The following information provides a brief overview of how conditions such as adrenoleukodystrophy (ALD) are added to newborn screening (NBS) panels in the United States, the United Kingdom, and in some European countries. Using the identified countries as examples, **the goal of this section is to give you a basic understanding of the required steps to add new diseases to a country's NBS panels and to obtain the necessary resources to finance implementation of the screening.** Once you have this understanding, you can start thinking about how to direct your advocacy efforts to advance ALD screening in your country or region.

This section may feel more technical and process-oriented than other components of this tool kit, mainly due to the need to refer to country-specific laws and regulations, but it includes relevant information for you to advocate more effectively. Each country-specific one-pager is divided into 3 areas of focus:

- 1 who are the **key stakeholders** that you will need to convince and who are the very final decision-makers that determine and decide whether to add a new disease to an NBS panel and to appropriate the specific expense for screening implementation
- 2 what is the **official process** and what are the required steps outlined in laws and regulations that will have to happen for authorities to effectively start screening for new diseases and for your goal to be considered and implemented;
- 3 what are the **criteria and evidence** that you will need to prove and present to have your request approved and to obtain a positive outcome

All this information can be a resource for you as you prepare for your engagements and give you a better overview of the data you might need to advocate effectively while navigating legal frameworks and conversing with stakeholders.



Don't forget, patient advocacy organizations can offer support as you begin your advocacy efforts in your specific state or country. Consider contacting an advocacy organization in your area. A list of resources including patient advocacy organizations can be found in section 7.



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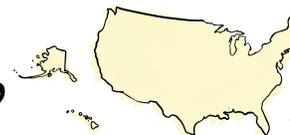
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The United States

Key stakeholders

In the United States, newborn screening (NBS) programs are managed at a state level. **Key stakeholders include the state's Department of Public Health, the NBS advisory committee (if the state has one), and any institutional review boards that may be involved in deciding which diseases might be added to their NBS panel.** There are additional federal agencies and programs that help states to implement and expand NBS programs.

- The Centers for Disease Control and Prevention (CDC) helps to ensure the quality and accuracy of NBS tests
- The US National Institutes of Health (NIH) invests in the identification, diagnosis, and treatment of conditions that could be appropriate for NBS
- The Health Resources and Services Administration disburses Title V Maternal and Child Health Services Block Grants, which provide funding for screening, treatment, and follow-up care

Despite federal contributions, states finance their NBS operations by appropriating state funding and often by charging fees to state programs and/or commercial health insurers to cover the laboratory costs associated with NBS.

The official process

The process for getting a disorder included on a state's NBS program in the United States usually begins with an application to the federal **Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)**. This committee bases its decision on several factors, such as:

- The condition being nominated
- Evidence that supports the potential net benefit of screening
- Availability of effective treatments
- Ability of NBS programs to check for the condition

If the committee deems these conditions to be met, it can make a recommendation to the Department of Health and Human Services (HHS) to add the considered disorder to the list that makes up the Recommended Uniform Screening Panel.

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Criteria and evidence

To qualify for the Recommended Uniform Screening Panel (RUSP), the disease or condition must meet 3 criteria:

- Condition can be identified 24–48 hours after birth (before clinical symptoms are present)
- A specific and reasonable test is available for it
- There are measurable benefits for early detection, intervention, and treatment of the disease

Unfortunately, not all states are required to screen for all of the diseases included on the RUSP. Newborn screening (NBS) programs in the United States vary between states due to several factors, including:

- Laws of the state
- Financial costs of screening and funding sources
- Risk and frequency of the condition in the state
- Availability of treatments and follow-up for each condition

In some states, the state law requires that the state's NBS program align with the RUSP within a certain time period after the addition of a new disease. In others, final decisions as to which conditions are coded on each state's panel are made by the state's public health department or advisory panel.

At that point, disease advocacy organizations, affected families, physicians, and biopharmaceutical companies are able to lobby each state individually to include the diseases in their state panel and to appropriate funding for screening implementation. To accelerate this process, there are a variety of steps individuals and advocacy groups can take:



Thanks to the efforts of advocates across the country, on February 16, 2016, adrenoleukodystrophy was added to the RUSP, the federal list of genetic diseases recommended for state NBS programs.



ORGANIZE

Establishing a larger organization of local groups can help raise awareness among the public as well as in the state legislature, especially if there is no presence at the national level.



PETITION

Address the ACHDNC directly. Even if they reject a new test, going through the process can raise awareness and deliver valuable support for future inclusion.



FUNDRAISE

Identify sources that are available, both federal and commercial. Getting funding from patient advocacy groups, drug companies, and governmental agencies for test development can speed the adoption of new tests while streamlining the process

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United Kingdom

Key stakeholders

In the UK, **policy recommendations on screening programs are made on a countrywide level by the UK National Screening Committee (NSC)**. The UK NSC was established to move screening policy away from a localized approach, with the aim to have all of the devolved constituent countries (Scotland, Wales, England, and Northern Ireland) follow the UK NSC's advice. The UK NSC meets privately 3 times a year, and its members include individuals with several different areas of expertise, such as health economics, pediatrics, laboratory services, and epidemiology, with cancer and public health being the most represented areas of expertise. The UK NSC does not involve rare disease patients in their work or review process; stakeholders may, however, be included in the evidence review process of an application by providing comments to the final review reports.

The official process

In the UK, the process to add a disease to the newborn screening (NBS) panel could either be **automatic**, ie, prompted by a regular review of current recommendations by the UK NSC committee itself, or through the **submission of a proposal** either to:

- 1 suggest a modification to an existing screening program
- 2 request an early update for a topic
- 3 propose a new topic that has not been previously reviewed by the UK NSC

The UK NSC conducts an annual call for new topics, starting in the first week of September and lasting for 3 months. Any individual or organization can submit a topic for consideration as long as they meet the UK NSC's application requirements. Indeed, each proposal will need to be submitted using the templated submission form to be found within the UK NSC website.

Using the submission form, the applicant will need to **explain** why the topic is within the remit of the UK NSC, **summarize and provide necessary justifications** as to why the criteria for adding a disease to the panel have been met, and also **provide up to 10 references** to support the application. All provided information should be based on referenced evidence published in peer-reviewed journals and be, ultimately, aimed at demonstrating that if a baby should be diagnosed with a condition through the NBS program, it would be able to access an effective intervention, be able to largely prevent or avoid the harm of the condition, with clear and extensive evidence that intervention at a presymptomatic phase would lead to better outcomes for the screened individual compared with usual care.

Criteria and evidence

The UK NSC criteria for appraising the viability, effectiveness, and appropriateness of a screening program are based on the criteria developed by **Wilson and Jungner** in the mid-1960s and relate to the **condition**, the **test**, the **treatment**, the **screening program**, and relevant implementation criteria.

The UK NSC will review the submitted application using an evidence review process. In particular, UK NSC will only consider evidence published in peer-reviewed journals and mostly, if not exclusively, literature or papers referencing UK-specific data. So far, only three types of evidence have been regarded as sufficiently strong to use as the basis for making recommendations: systematic reviews, randomized controlled trials, and population-based studies.



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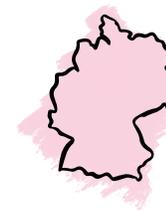
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Germany

Key stakeholders

The decision to include a new disease in the newborn screening (NBS) program and into the reimbursement catalogue of the statutory health insurances lies with the **Federal Joint Committee (G-BA), the Institute for Quality and Efficiency in Health Care (IQWiG)**, and, ultimately, with the **Ministry of Health**. The official process starts with a **member of the G-BA plenary petitioning** for the assessment of a new “screening” method. The G-BA plenary comprises 13 members, from statutory health insurers and healthcare providers to impartial participants. In particular, except for the impartial participants, the members of the G-BA plenary are part of the following organizations: the Umbrella Organization of Statutory Health Insurers (**GKV-Spitzenverband**), the Association of Statutory Health Insurance Physicians (**Kassenärztliche Bundesvereinigung, KBV**), the Association of Statutory Health Insurance Dentists (**Kassenzahnärztliche Bundesvereinigung, KZBV**), and the German Hospital Association (**Deutsche Krankenhausgesellschaft, DKG**). Patient advocacy organizations are nonvoting members and only have the right to petition and co-consultation.

The official process

Contrary to what other countries do with respect to extension of the NBS programs, the G-BA does not assess a disease to be included in the screening catalogue, but considers a new screening as **a new “method” or “examination.”** In particular, in accordance with §135 of the 5th Book of the German Social Code (SGB V), the G-BA is mandated to assess the **therapeutic benefit, medical necessity, and cost effectiveness** of new examinations and/or treatment methods. Based on the result of this review, the G-BA decides whether a new examination for the early detection of illnesses should

be eligible for reimbursement by the statutory health insurance. Considering that the official process starts with a petition from G-BA, it needs to be stressed that patient advocacy groups and healthcare providers may suggest to the G-BA a disease for inclusion in the NBS catalogue by **submitting a proposal**.

After an initial consultation, the G-BA mandates the **IQWiG to conduct an assessment**, which, apart from the relevant scientific and economic analysis, will also include input from several stakeholder groups, including the commission for gene diagnostics at the Robert-Koch-Institute. Indeed, medical societies and patient advocacy groups may provide their own opinion by completing a **questionnaire**, which will be taken into account by the IQWiG when it drafts its final assessment report. Based on the final IQWiG report, the G-BA will hold an expert hearing and could recommend the reimbursement of the screening procedure by the federal health insurances. Unless the Minister of Health objects to the G-BA’s recommendation, the recommendation enters into force 1 day after the Ministry’s publication in the Federal Gazette.



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France

Key stakeholders

As a result of the administration's endorsement of a public action on newborn screening (NBS), starting in 2018, the French NBS program was completely reorganized. As part of this new organization, the Minister of Health announced the establishment of a **National Steering Committee, chaired by the Director General of Health**, whose role would include "to propose to the Minister of Health any change in the list of detected diseases." The National Steering Committee would include representatives from the following agencies: the Directorate General of Health (DGS), the Directorate General for the Provision of Care (DGOS), the Superior Health Authority (HAS), the Agency for Biomedicine (ABM), the National Agency for the Security of Medicines and Health Products (ANSM), the National Agency for Public Health (ANSP), ARS, CNAMTS, CCNE, CRDN, SFSP, the French Pediatric Society (SFP), and the French NBS Society (SFDN).

The official process

The current process to add a disease to the NBS panel requires that the National Steering Committee propose modifications of the list of diseases **directly to the Minister of Health, who will change the list of diseases by issuing a binding order, an "arrêté."**

It is important to highlight that the National Screening Committee's proposal shall be made only after the occurrence of a **mandatory screening assessment by HAS**, with the collaboration of the ABM if required.

Several questions, however, remain unanswered. Who triggers the HAS assessment? What is the division of roles among the HAS, the ABM, and the National Screening Committee? What is the timeline of the procedure and the information required?

The HAS previously affirmed that it will assess the value of including approximately 30 diseases, as it is assessing the feasibility of extending the NBS panel to metabolic diseases using the mass tandem spectrometry technique.

Considering the multitude of organizations represented within the National Screening Committee and the fact that the proposal will need to come from the Committee itself, it is clear that patient associations and/or scientific societies will be critical for raising awareness and highlighting community and scientific consensus regarding the importance to add a specific disease to the NBS program.

Criteria and evidence

The criteria that are considered for the French NBS Program are those that lay the foundation for the majority of NBS programs. Indeed, according to the **Wilson-Jungner criteria**, the disease must be **serious**, have an **effective treatment**, and be detectable at a presymptomatic stage via a **simple, reproducible, and reliable test**. Notwithstanding, there is still a need to obtain clarity on the specific criteria utilized by the HAS in its preliminary assessment and by the National Steering Committee in deciding whether to submit a proposal.



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Italy

Key stakeholders

The National Agency for Regional Health Services (**AGENAS**), the Higher Health Institute “**Istituto Superiore di Sanità (ISS)**,” the newborn screening (NBS) Center of Coordination established within the ISS “**Centro di coordinamento per lo screening neonatale presso l’Istituto Superiore di Sanità**,” the sector-specific **scientific societies**, and the **Ministry of Health** will all play a role in adding a new disease to the Italian NBS panel. The need for overall scientific and political consensus is therefore specifically important in Italy considering how the Ministry of Health will need to collaborate and consult with several institutions before the final decision is made to add a disease to the national NBS panel.

The official process

The official decision-making process to add a disease onto the Italian NBS panel is detailed in the Italian NBS legislation, also called “**Taverna law**” (L.167/2016), as recently amended by the 2018 budget law.

The process requires the Ministry of Health, in collaboration with the ISS, AGENAS, the regions, and the autonomous provinces of Trento and Bolzano **to review the current list** of screened pathologies **every 2 years** on the **basis of ongoing progress of scientific evidence** in the relevant diagnostic and therapeutic fields for hereditary genetic diseases. The Ministry’s decision will also depend on **mandatory consultations** with relevant scientific societies, and, based on a systematic interpretation of the law, on the results **of health technology assessment evaluation** to be carried out by AGENAS.

Even if the decision-making process has been recently detailed in legislation, some doubts remain with respect to the relevant “application” process. The Ministry may update the list of screening pathologies based on informal requests coming from patient associations and/or scientific societies and brought forward by the NBS Center of Coordination.

Criteria and evidence

In order to be potentially successful, all relevant requests should be able to demonstrate the fulfillment of the following criteria: (i) **severity of the illness**, (ii) existence of an **efficient laboratory test** selected on the grounds of its diagnostic accuracy in differentiating potentially affected subjects from the normal population, and (iii) availability of an **effective treatment** able to substantially modify the natural history of the illness, if adopted early. These criteria have been used to compile the current list of approximately 45 diseases that are included into the extended NBS program, together with (iv) the comparison with the recommendations provided within the “Guidelines for the extended neonatal screening and for diagnostic confirmation” published by the Italian NBS scientific society, and (v) comparison with international experiences.

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The Netherlands

Key stakeholders

The stakeholders who will ultimately make the decision of adding a new disease to the newborn screening (NBS) panel in the Netherlands are the **Health Council of the Netherlands (Gezondheidsraad)**, the **National Institute for Public Health and Environment (RIVM)**, and the **Minister of Health, Welfare, and Sports**. It is interesting to note that, in the Netherlands, public health falls within the responsibility of the State Secretary.

The official process

The process starts with an **advisement by the Health Council** of the Netherlands (Gezondheidsraad) to expand the NBS panel. The advice coming from the Health Council automatically prompts the National Institute for Public Health and Environment (RIVM) to start a so-called **feasibility study** to see whether the expansion of the NBS panel may be needed or is otherwise feasible.

Based on the results of the RIVM study, the Minister of Health makes a preliminary decision. The Minister may either **opt for a phased implementation** or directly **make a final decision**. As it relates to the phased implementation, it is usually based on phased pilot projects, including various validation tests managed by an academic research team. The phased implementation is supervised by the RIVM. Based on the results of the pilot, the minister will make a final decision on the addition of the disease to the NBS panel, including the timing of the addition.

Criteria and evidence

The Gezondheidsraad's advice and final recommendation to extend the NBS panel is based on the following 5 criteria: (i) **improved scientific knowledge**; (ii) the screening should be aimed at **preventing irreparable damage** to the child and/or achieving significant health benefits for the child; (iii) the screening should be able to permit the **shortening of the diagnostic process**; (iv) the screening should be aimed at **facilitating reproductive choices** for parents; and (v) the screening should help to **alleviate the burden** of disease in society.



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“The time to act is now. A validated and accurate screening test for adrenoleukodystrophy is available. We highly recommend adding newborn screening for ALD to the current state panel. Let’s do what we can to help young boys avoid suffering the devastating effects of this deadly disease.”

-BRAD ZAKES

ADVOCATE IN WASHINGTON

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Importance of framing

Every story has a beginning, middle, and end. Each piece of the message matters. To ensure that your story has the most impact for your audience, you'll want to frame it with a strong beginning that captures attention and a memorable ending that calls the listener to action.

Start with a strong statement that lets your audience know you recognize that your voice matters. Have confidence in what you have to share and the importance of your message.

Consider beginning with a statement of the personal impact of newborn screening (NBS), or lack thereof, on you, your family, or someone else you know who is affected by adrenoleukodystrophy (ALD). Remember, **you are the expert of your own story.**



Examples of strong initial statements from the parents and caregivers of children with ALD:

Example: *The day my son was diagnosed with ALD was too late; I was aware that he should have been screened for this. He could have been diagnosed earlier. We could still have our son with us today.*

Example: *We entered the hospital with a little boy full of energy and personality, and we left the hospital with a little warrior who couldn't walk and couldn't communicate with us. We were all just devastated. We needed to learn a new life. NBS could have prevented this outcome for our family.*

Example from Brad Zakes: *We are like so many other stories. We just thought our son had attention deficit disorder or that he was just kind of a unique little kid. But it wasn't until his symptoms got to the point where we knew there was something really serious going on that it was really too late. As horrific and difficult a diagnosis may be for a family, just knowing gives them the benefit of time, to be able to monitor the disease and be in a position where they can take necessary medical intervention at the appropriate time. That in my mind is the key. It will save lives. I think it already has.*

Example: *ALD doesn't just happen to just 1 boy. This is not an isolated event. The disease affects 1 in 21,000 males born each year. The devastation of this disease can be stopped and the cost to do so is minimal.*



End with a memorable action that gives your audience a way to get involved. The action you end with may encourage a stakeholder to become an advocate, inspire an influencer to share your message, or activate a decision-maker to promote NBS for ALD.

Example: *Will you help me to share my story and bring awareness of ALD and the importance of NBS to a larger community?*

Example: *How can we work together to ensure families receive early diagnosis of ALD and can properly monitor for disease progression before it's too late?*

Note: *The examples shown above are not real stories, just ways to illustrate the importance of framing.*



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ACTIVITY - DEVELOPING YOUR STORY

Try responding to each of the questions below in 1 or 2 succinct sentences. Then organize your responses into an order that makes sense for your overall message. You'll find your story beginning to develop. Don't forget to frame your story with a strong beginning and a memorable ending. And be sure to sprinkle in some facts about adrenoleukodystrophy (ALD) and newborn screening (NBS) to add to your message.

What is ALD?

How has ALD affected you and your family's life?

Why is early detection of ALD critical to you? To all those diagnosed with ALD?

How did NBS play a role in your family member's diagnosis, if at all?

Why do you think NBS is important and how has it affected your life?

What action do you want to leave your audience with?



Example from Brad Zakes: *In May 2011, Ethan passed away at the age of 10, just 5 months after being diagnosed with cerebral ALD. ALD is a genetically acquired degenerative neurological disorder that causes damage to the nerve cells of the brain and the adrenal glands. Approximately 1 in 21,000 boys are born with the condition each year. Unless a family history of ALD is known, the disease is rarely detected at birth. As a result, most young boys appear to be completely healthy during their early childhood and remain undiagnosed until later in life (at approximately 4 to 10 years of age), when they start to show initial symptoms—frequently too late to stop the progression of this devastating disease. As a result, death is an unfortunately common outcome for many young boys. NBS provides the opportunity to detect the disease shortly after birth. By simply detecting the disease early in life, boys diagnosed with ALD can be monitored for cerebral progression and receive treatment to better their chances of living a normal, healthy life. A highly reliable and accurate screening method utilizing dried blood spots from standard neonatal screening cards has been developed to diagnose ALD when it matters, early.*

Remember, it is likely that **most of the individuals** with whom you share your story will **have never heard of ALD**. Others may be unaware of NBS. This is your opportunity to educate. **You are the expert.** As you share your personal story with them, don't forget to answer the basics by defining the condition, the incidence, and the potentially devastating impact. Consider visiting [NavigatingALD](#), [ALD Connect](#), [ALD Alliance](#), or other patient advocacy organizations and resources for information and facts for you to include in your message.



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ALD and NBS facts and figures:

- Newborn screening (NBS) is a process in which **newborns are tested for a select group of rare diseases** to help prevent symptoms and allow for early treatment
- Adrenoleukodystrophy (ALD) is a rare, X-linked disease that occurs in about 1 in 21,000 males and about 1 in 17,000 newborns in the total population
- ALD is caused by an underlying genetic mutation in the *ABCD1* gene and affects the body's ability to create the protein that helps in the process of breaking down very long-chain fatty acids (VLCFAs)
- A buildup of VLCFAs can cause adrenal problems and can potentially lead to brain damage. **ALD that progresses** to affect the brain is referred to as **cerebral ALD**
- **About 40% of boys with ALD will develop cerebral ALD**, which leads to irreparable, progressive demyelination and neurodegeneration resulting in disabilities such as an inability to speak or respond, blindness, or even death

Diagnosis and treatment

- **Early diagnosis of ALD** and signs of cerebral demyelination and neurological symptoms can save lives. Once ALD is diagnosed, it is recommended that affected boys be monitored regularly with a brain MRI to detect cerebral involvement as soon as it develops, and treatment can be considered
- If treated early, **stem cell transplant can dramatically improve the outcomes** of affected boys

- **If treatment is delayed** until the condition is apparent clinically, **outcomes are shown to be worse**, frequently leading to death
- **NBS for ALD is the earliest form of detection** and is an opportunity for families to engage in monitoring for potential progression to cerebral ALD

NBS for ALD

- **NBS for ALD saves lives**
- Without NBS, early lifesaving therapy has been available only to those diagnosed because of diagnosis through known family history
- The **ALD NBS test is accurate and inexpensive**, with an estimated price of about \$5.00-\$11.00 per test in the United States; globally, this cost varies
- The screening method utilizes dried blood spots from standard neonatal screening cards
- The test is performed via high-throughput screening assays using tandem mass spectrometry
- The test has a high level of specificity (low false-positive rate)
- ALD met all of the criteria for inclusion on the United States' national Recommended Uniform Screening Panel and is currently screened for in 18 states as of January 2020



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Support letter template

When advocating for newborn screening (NBS) of adrenoleukodystrophy (ALD), it may be important to include a letter of support or two from a healthcare provider, institution, or expert from the leukodystrophy community who can verify and bolster your message that NBS for ALD is critical. To the right is a template that you can use to adapt a letter of support with a provider, institution, or leader of your choosing whose voice can help to support your advocacy efforts.



Request the letter be placed on official letterhead from the healthcare provider, institution, or opinion leader you are working with to adapt the letter. This can offer some clout and additional authority to your overall message.

Remember, advocacy organizations may be able to help you to identify someone to sign on to your letter of support.

You can create your own letter by zooming in on the template below and copying language to help get you started. From there, add your own individual touches to make it feel more personal.

First Name Last Name mm/dd/year
 Organization, Title
 Address
 City, State Zip Code, Country

Dear Mr./Mrs./Ms. Last Name,

I am writing to express strong support for the implementation of newborn screening (NBS) for adrenoleukodystrophy (ALD), an X-linked genetic disorder that occurs in about 1 in 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 in the process of breaking down very long-chain fatty acids (VLCFAs). A buildup of VLCFAs can cause adrenal problems and can potentially lead to brain damage in a form known as cerebral ALD. Unfortunately, many boys with ALD go undiagnosed until symptoms of cerebral disease have developed. If cerebral ALD is left untreated, there are severe side effects, including significant disabilities, such as an inability to speak or respond, blindness, or even death.

Hematopoietic stem cell transplantation with related and unrelated donors can prevent progression of the cerebral form of ALD if performed before or soon after the patient has manifested clinical symptoms of the disease. If diagnosed early, MRI may be able to detect the advancement to cerebral disease before clinical symptoms appear and at a time when treatment may be effective. Conversely, if transplantation is performed in symptomatic children, the disease usually progresses and the child either dies or stabilizes in a severely debilitated state.

Most families are unaware of the fact that they are at risk for having a child with ALD until the child becomes symptomatic. Usually it's too late to help at this time. Although subsequent pregnancies or births can be screened, this approach fails to rescue the first child who could have been diagnosed early, and could have had access to a hematopoietic stem cell transplant, but was not.

Another important reason to identify these children with NBS is because some will present with life-threatening problems due to adrenal crises, which arise as adrenal insufficiency develops. In many children this occurs before the onset of neurological symptoms. Given that adrenal crisis is also life-threatening, it is a second compelling reason to screen newborns for ALD.

NBS is an intervention that offers the potential to save the lives of hundreds of children with ALD. Early diagnosis of ALD followed by routine, serial brain MRI screening has the potential to dramatically impact treatment options for affected boys with ALD. The critical aspect in achieving optimal outcomes is detecting the disease before it results in too much damage to the brain. In this circumstance, NBS is an excellent candidate for making a difference in this devastating disorder.

ALD was added to the Recommended Uniform Screening Panel in the United States in 2016, verifying that NBS is the recommended diagnostic option for ALD. With a sensitive, specific, and timely test developed and able to be adapted for mass screening, the overtime and overall cost-effective nature of screening for ALD at birth cannot be argued.

In summary, I highly recommend that <<Insert Decision-Maker>> undertake the task of adding NBS for ALD to the current panel. Please do not hesitate to contact me if you require additional information in support of this request.

Sincerely,
 <<Insert Signature>>
 <<Insert Contact Information>>



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Encourage advocacy from others— sharing your message with stakeholders

It's no secret that more voices can expedite and enhance your overall message: **newborn screening (NBS) for adrenoleukodystrophy (ALD) is necessary!**

Consider sharing your story with other ALD families and broader community members to communicate the importance of NBS for ALD and the impact their voices can have with yours.

Reach out to one or more patient advocacy organizations to connect with other families in your area. Invite them to join you in advocating. Advocacy organizations can help you to connect to other advocates or families in your region and assist you in navigating the advocacy process, identifying decision-makers, and sharing your message.



“Connect with the people who have been involved in this community and with advocacy. Talk to them. Ask them questions. Families who have been dealing with this longer are truly some of the greatest resources. I wouldn't be the advocate I am without them. Connect with the community. Don't be afraid to seek help and ask questions just because you feel you may not know enough about the disease or how the process works.”

– FRANI BROUSSARD / advocate in Texas

Section 7 of this tool kit includes a variety of resources, such as patient advocacy organizations you may choose to contact that are active in ALD and NBS.



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Contact your local media outlets: Sharing your message with reporters/influencers

The media can be a partner to you in sharing your story and the importance of newborn screening (NBS) for adrenoleukodystrophy (ALD). Consider approaching a variety of media outlets, including community newsletters, local newspapers, regional news agencies, etc.

Parents Lobby States to Expand NBS Test for Rare Brain Disorder

When connecting with media outlets, consider the following questions:

- Who is their audience?
- How wide is their reach?
- How can you gain their attention?

Knowing the answers to these questions will help you to prepare your message and method of contact.



I pretty much reached out to the major news channels with my story to try to get them interested in raising awareness about ALD and about NBS. My suggestion to others would be to reach out to your local newspaper and television stations and get coverage that way. Let them know your story and the story of NBS.”

– JANIS SHERWOOD / advocate in California

Janis shared her story with local media as she advocated for ALD NBS in California. Here is an article that was printed in the San Diego Union Tribune about her advocacy efforts to raise awareness of ALD, courtesy of the San Diego Union Tribune:

Janis and Sawyer’s story



You might be surprised how connected you and your immediate network may be. Advocate Frani suggests sharing your efforts broadly and talking about them often.

“I tell everyone I know; sometimes you may not want to talk about it, or it feels like you are burdening someone with what is affecting your life. But the more you share the more you find that people are truly willing to help. Talking to a network is important. People want to help. They want to help you and they want to help your family.”

When working with the media:

- You can ask to see questions in advance of potential interviews
- You can suggest questions in advance of potential interviews
- You can rule out topics you prefer not to respond to



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Encourage change through action: Sharing your message with decision-makers

Once you have identified the decision-makers, you will want to prepare information to help them to see the critical importance of newborn screening (NBS) for adrenoleukodystrophy (ALD).



Change comes from the action of sharing your story; this is Jean Kelley’s message about the importance of advocacy.

“The most important thing we did was make an appointment with the Health Committee in Connecticut. I made some fact sheets and took Brian with me and explained to them what ALD is and what Brian’s challenges had been since he was diagnosed and all that he had been through. We shared that there was an opportunity for NBS to identify this disease early and help children to survive without the hardships that Brian had with early detection, monitoring, and treatment.

The impressive thing was that Brian sat quietly and listened and moved his eyes to those who were speaking, although he doesn’t see. And he moved his lips to answer questions about things that he liked... just yes or no questions, but he was able to communicate. They could understand that he was convinced that no one should have to live like that. They realized that if they could do something about this disease, they should. The committee approved the bill and efforts started to move forward. I impressed upon them that NBS is money-saving and how expensive is Brian’s schooling and health care, and how fortunate we were to have private insurance, whereas a few other families in the state were dependent on state healthcare.”



I encourage other advocates to share the power of their experience with ALD, and also to touch upon the money-saving aspect of NBS. It’s the bottom line. It went through appropriations, so they have to see things from that perspective. We invited our representatives to our home, let them in and showed them everything we need to accommodate Brian’s new needs—a lift, a ramp, a specialized bathroom—all of the things that made Brian’s life easier for him and for us.”

– JEAN KELLEY / advocate in Connecticut



As an advocate, consider using your child’s medical bills as a representation of just how costly diagnosis outside of NBS can be on a family and on a healthcare system.

Please only contribute personal information that you are comfortable with sharing.



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Bring a leave behind

Many advocates choose to bring a small package of information such as a leaflet with them when speaking with decision-makers to support their overall message.

This package of information may include:

- **Your story (1 page)**
- **A few images of individuals affected by adrenoleukodystrophy (ALD) (approximately 3 to 5)**
- **Quick facts about ALD and NBS (1 page)**
- **Letter of support (approximately 2 to 3)**
- **Contact information (2 business cards)**

Consider packaging your leave-behind in an open folder to give to a decision-maker. Some offices will not allow materials to be left behind in closed envelopes for security reasons.



You may want to include the contact information for a local Patient Advocacy Organization with expertise in leukodystrophy and/or newborn screening (NBS) within your materials. This will allow individuals another outlet to reach out to with questions. You can start to identify patient advocacy organizations in the resources section of this tool kit.



I always bring a one-sheet of ‘What is ALD’ with basic facts. As you can imagine, all of the legislators are busy and aren’t going to become experts in this disease. We need to provide them information in an easily accessible way. I also bring letters of support from institutions and clinicians and physicians who also advocate that newborn screening is necessary. I then leave these pieces behind with them. If they have questions, they have something to go back to. Telling your story is the most important thing, but then leave those fact sheets behind as a reminder.”

– ELISA SEEGER / advocate in New York

Make an appointment to meet with decision-makers in person or via video. Bring your package of information with you and be prepared to speak with members of their staff before ever having an opportunity to speak with the decision-maker directly.



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How will you share your story?

With today's technology, you may be contacting stakeholders, influencers, and decision-makers in a variety of ways:



EMAIL



FACE TO FACE



PHONE



MEDIA
(PRINT,
DIGITAL,
TV)



SOCIAL
MEDIA/ONLINE
DIGITAL
PLATFORMS

At times, you may be engaging in cold-calling or cold-emailing by contacting individuals you do not know but who you have identified as a key stakeholder, influencer, or decision-maker in your region. You might identify these individuals by the role that they serve, the area of focus they work in, the causes they have supported previously, or the influence they hold in your community

“ You get more ‘no’s’ than you get ‘yeses. Don’t be discouraged. When you get that one ‘yes’ all of the ‘no’s’ disappear. How many kids can that one ‘yes’ save? I always try to stay focused on...if my efforts to raise awareness of adrenoleukodystrophy and newborn screening saves just 1 boy, that’s a life... that’s worth it. Don’t give up. Sometimes it takes multiple attempts before someone will hear you out on what you have to say.”

- JANIS SHERWOOD / advocate in California

“ I find people to be receptive to connecting and hearing me out. Advocates should always be proactive, anticipate someone’s questions, and leave the door open for someone to get back to you when they’re ready to learn more... I send a lot of emails and make a lot of phone calls. I often find people through social media.”

- BOB WYBORN / advocate in Australia



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Tips for cold calling

- In advance of your call, **research** the individual you are calling and note why you have chosen to reach out to them about newborn screening (NBS) for adrenoleukodystrophy (ALD)
- **Open with a strong statement** that shares who you are and why you are calling
- Do not ask “Is now a good time?” It’s always a good time to advocate and educate. **Launch** straight into your purpose
- **Describe ALD** and the critical importance of early diagnosis
- **Let the listener know** that NBS for ALD is already being implemented in some states in the US and that a test for ALD NBS is already available and might only need to be localized
- **Make an ask.** Prior to your call, predetermine what action you want the listener to take at the end of the conversation. You may want to set up a meeting. You may want to get their ‘OK’ to do a news article on your family. You may want to get their ideas for how to raise awareness on social media
- Whatever your ask is, don’t end the call until you’ve **shared an action**
 - Ask for an appointment at a specific time. You want your listener to commit to a meeting so you can share more
 - Ask for the name and number for a colleague who focuses on human interest pieces
 - Ask what additional information they may need to consider advocating with you for NBS for ALD



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Take your voice on social media

Social media can be a powerful tool for advocacy efforts and allow you to educate and inspire a broad audience. There are more than 3 billion Internet users around the world, with more than 2 billion people having a social media account. With so many different social media platforms at our fingertips, it might feel overwhelming to decide which platform to use and how to best share your message. There is no correct answer, but Twitter is a platform where key decision-makers tend to engage more actively.



Each September is newborn screening (NBS) Awareness Month. Help to raise awareness of the importance of NBS through hashtags, such as:

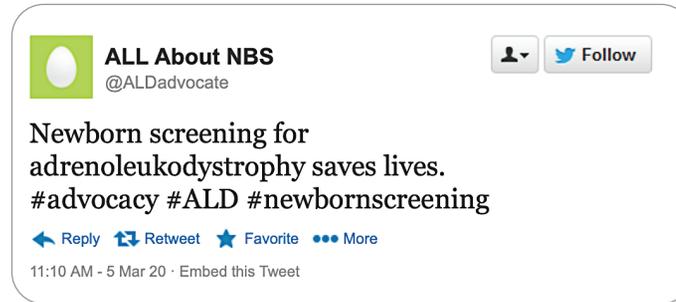
#NewbornScreeningAwareness #NBS
#NewbornScreeningMonth #newborntesting
#NBSMatters #NBSSavesLives

Twitter

Members of Congress and Parliament, legislative staffers, and policy makers tend to have an active presence on Twitter. As a platform, Twitter allows users to have real-time conversations. Consider using hashtags such as **#ALD**, **#adrenoleukodystrophy**, **#raredisease**, **#newbornscreening**, **#advocacy**, and **#myvoice** to broaden your message's reach.

Remember: Keep your social message short and sweet, 280 characters or less. Your tweet should galvanize your audience to action and/or heightened awareness. Remember that tweets with photos are more likely to be shared and will help spread your message.

Example tweet:



Tip: Identify which representatives you want your message to reach on Twitter and be sure to follow their chief of staff, legislative assistant, and close network. You'll want to engage with their posts to ensure that when you share your messages, they'll show up in the right places to make an impact.

Social Media Accounts Related to NBS:

You can amplify your message and those of others around NBS by following and engaging with accounts that are already raising awareness for the cause. Consider following **@EveryLifeOrg**, **@BabysFirstTest**, **@GlobalGenes**, **@RareDiseases**, or **@Eurordis**.



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“ I always bring a one-sheet of ‘What is ALD’ with basic facts. As you can imagine, all of the legislators are busy and aren’t going to become experts in this disease. We need to provide them information in an easily accessible way. If they have questions, they have something to go back to. ”

- ELISA SEEGER
ADVOCATE IN NEW YORK

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Adrenoleukodystrophy and leukodystrophy patient advocacy organizations*

- [AIALD](#)
- [ALD Alliance](#)
- [ALD Connect](#)
- [Brian's Hope](#)
- [Calliope Joy Foundation](#)
- [Ethan Zakes Foundation](#)
- [Fight ALD](#)
- [Global Leukodystrophy Initiative](#)
- [Leukodystrophy Resource and Research Organization](#)
- [Hunter's Hope](#)
- [United Leukodystrophy Foundation](#)

Newborn screening (NBS) and rare disease advocacy organizations

- [AISMME](#)
- [Baby's First Test](#)
- [Everylife Foundation](#)
- [EURORDIS](#)
- [Global Genes](#)
- [IPOPI](#)
- [National Organization for Rare Disorders](#)
- [UNIAMO](#)

General advocacy resources

[A guide to public speaking](#)

NBS resources

[What is NBS? Animated video for parents](#)

[The role of laboratories in NBS](#)

[Example of NBS blood specimen collection](#)

[Example of economic impact of NBS for ALD](#)

[Advocacy for the community NBS](#)

[Importance of NBS](#)

Rare disease resources

[Using storytelling to raise awareness for your rare disease](#)

[How to promote your rare disease story through social media](#)

[Contact a representative \(US\)](#)

Note that this is only a sample listing of resources that may be helpful to you in your advocacy efforts.



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“ 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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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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