4 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

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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**

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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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.

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.

> "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 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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