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April 6, 2017

Senate Health Committee
Chair – Sen. Laurie Monnes Anderson
Vice-chair – Sen. Jeff Kruse
Members – Sen. Lee Beyer, Sen. Elizabeth Steiner Hayward, Sen. Tim Knopp

Re: SB 808 – Newborn Screening

Dear Chairperson Monnes Anderson and the Health Committee,

I regret that I am unable to join your today to give this testimony in person. I am a rare disease dad and have medical appointments for my daughter that conflict with this hearing. I was unable to reschedule due to a very busy travel schedule. I am on a flight home as I write this – completing my 4th 2017 trip to Washington DC working on research and policy.

Two of my three daughters have a very rare disease called Metachromatic Leukodystrophy where the insulation on their nerves degrades resulting in shorted out nerve signals. The outcome of MLD is terminal when children are diagnosed after symptoms occur. It took us 6 years to get to a diagnosis based on improper assessment of clinical symptoms. Newborn screening would have enabled us to seek treatments and therapies before disease progression and would have allowed my daughters to live a full productive pain-free life.

In addition to being a rare disease parent, I am also co-founder and unpaid/volunteer President of MLD Foundation where we have served families and impacted researchers, academic institutions, clinical facilities/hospitals, and policy makers worldwide since 1999.

I realized early on that our MLD process and policy issues are very similar to the 7,000 other rare diseases when it comes to policy so I invest a lot of time in Rare Disease policy. I have been aggressively involved in Newborn Screening at the federal

and state level for 7 or 8 years and have attended most of the ACHDNC[1] (HHS Secretary's Advisory Committee for Heritable Disease in Newborns and Children) during that time period. While the ACHDNC's recommendations are only guidelines and cannot force states to implement screens, they are designed to make sure a river or barb wire fence between states is not the difference between a baby receiving a life saving screen or not.

I was also the inspiration for the focus of California's 2016 SB 1095[2] which inspired SB 808 in Oregon. Please note that neither of these bills is disease specific and that MLD does not have an application in front of the ACHDNC – these are Rare Policy issues addressing the needs of the 1 in 10 of us that are born with a rare disease – truly a public health focus.

I have the greatest respect for the extensive evidence-based review the federal AHDNC Committee performs before recommending a new screen be added to the RUSP (Recommended Uniform Screening Panel). The Committee is made up of a broad group of members including public health at the state and federal level, newborn screening (NBS) lab directors, clinicians, and a variety of other expertises/ perspectives.[3] They use an external review panel made up of research and clinical experts for a very scientific evidence-based 6-month review of every new application.

The 11 primary criteria for a RUSP recommendation are based on criteria established by the ACMG (American College of Medical Genetics) in 2003 which has its roots in the 1968 Wilson-Jungner criteria.[4] [5] The primary criteria include:

- An accurate repeatable newborn screen
- A requirement that timely identification of the disease at birth changes the child's medical care resulting in a benefit to the child
- Availability of an accurate conforming diagnostic test
- Availability of a viable therapy to help those identified with a disease
- The screen is implementable in state labs, including being cost effective, efficient, and practical in terms of methodologies. Usually this is demonstrated by reporting on a Pilot Study where the NBS is implemented in 1 or more states for 1-2 years to gather data on the screening of 10's if not 100's of thousands of newborns.

The RUSP application is thoroughly reviewed and scored. Only those screens meeting minimum individual and total standards are sent to the HHS Secretary for review and consideration. The HHS secretary typically engages additional outside review by the CDC and other resources. This review process usually takes several years. The initial application may be preliminarily reviewed and rejected by the

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[1] https://leginfo.legislature.ca.gov/faces/billNavClient.xhtml?bill_id=201520160SB1095

[2] https://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/about/memberroster.pdf

[3] https://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/index.html

[4] http://www.nature.com/gim/journal/v15/n3/fig_tab/gim2012176t1.html

[5] http://www.who.int/bulletin/volumes/86/4/07-050112/en/

committee before external review for not being complete and it may be rejected by the external review committee based on the scientific evidence before being subjected to a final review. And then, often one or more of the 11 criteria is not adequate for the Committee to make a RUSP recommendation. There are currently 34 screens on the primary RUSP panel.[6] Oregon does not currently screen for all 34 recommended conditions so a baby born Vancouver WA, Yreka CA, or Tampa ID may get a life-saving screen that one a few miles away in Oregon will not.

Developing a screen, running a pilot study, preparing a RUSP application, ACHDNC review, and HHS Secretary review often takes 5+ years. But with that said, the real journey begins after the condition is added to the RUSP. That is when the state labs start their implementation.

Newborn screening is much more than the laboratory analysis of a blood spot. NBS is part of the state public health programs. Processes for how to get to a definitive confirming diagnosis after a potential-positive indications from a NBS screen, contact and follow-up procedures with families, and how to uniformly implement the screen across the state so all families have access are some of the broader considerations when implementing a screen. This can take years … historically 4 years or more. The gating implementation issue is usually the state lab's implementation of processing and the analytic capability of the screening test.

SB 808 focuses on directing the Oregon NBS lab, which services Oregon and several other sates through a consortium, to become more structured, timely, and predictable with regard to implementing new screens. It establishes a two year implementation deadline based on the later of being added to the RUSP or the availability of a FDA approved screening kit for a RUSP condition. It is important to note this state lab clock does not start suddenly. The lab has upwards of five year's notice as they monitor applications for RUSP approval proceed through the federal review process, all the while providing feedback to their representatives on the federal committee. And remember that part of the RUSP application is a successful Pilot Study in one or more labs that successfully demonstrates the new screen's ability to be implemented and its effectiveness.

SB 808 also allows the lab the flexibility to develop its own Laboratory Developed Test (LDT) when it feels they may have a better or more efficient screening methodology. This deadline is 3 years due to the extra challenges of a LDT

SB 808 also establishes a legislatively mandated Newborn Screening Committee with a charter to engage various perspectives, including citizens, into a two year process to review and discuss challenges, limitations and opportunities for the state NBS lab … all to be reported back for the 2019 legislative session. We want Oregon's NBS program to be a leader in the nation just as we are innovative leaders in other aspects of our public health system.

The Oregon Department of Health and the Oregon Lab have offered to convene such a committee without legislative mandate … and they have indicated that a two year

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[6] https://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendedpanel/index.html

implementation deadline may be challenging – even with 5 years impending notice. During this legislative process the past few months we have built solid relationships with Department of Health and Lab personnel and we highly respect their skills, perspective, and intentions. None of us like more structure, but we believe it is in the best interest in the 1 in 10 of us born with a rare disease to have some strict time and activity deadlines for test development and reporting back to the Senate about our newborn screening public health program.

I have agreed that a strong show of good faith by the Health and Lab teams to support the SB 808 timelines for implementation of new tests based on RUSP approval would cause me to support the removal of the legislatively mandated committee in favor of a commitment to meet in a slightly less formal manner and still report back to the 2019 Senate.

Sincerely,

Dean Suhr, Oregon Rare Disease Dad
President, MLD Foundation

cc: Sponsoring Senators Boquist and Beyer
Sponsoring Representatives Buehler and Parrish

Co-Sponsoring Senators Devlin, Kruse, Monnes Anderson, Olsen, Roblan, Thomsen
Co-Sponsoring Representatives Barnhart, Heard, Kennemer, Keny-Guyer, Lively, Malstrom, Nosse, Witt

Table 1: Wilson and Jungner classic screening criteria

1.	The condition sought should be an important health problem.
2.	There should be an accepted treatment for patients with recognized disease.
3.	Facilities for diagnosis and treatment should be available.
4.	There should be a recognizable latent or early symptomatic stage.
5.	There should be a suitable test or examination.
6.	The test should be acceptable to the population.
7.	The natural history of the condition, including development from latent to declared disease, should be adequately understood.
8.	There should be an agreed policy on whom to treat as patients.
9.	The cost of case finding (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care as a whole.
10.	Case finding should be a continuing process and not a "once and for all" project.

Data from ref. 22.

http://www.nature.com/gim/journal/v15/n3/fig_tab/gim2012176t1.html