



SOUTH DAKOTA DEPARTMENT OF HEALTH

Every South Dakotan Healthy and Strong

South Dakota Newborn Screening Advisory Committee

April 28, 2022
 10:00am – 12:00pm CT
 Tieszen Building – Basement Level Conference Room and Zoom
 306 E Capitol Ave.
 Pierre, SD 57501

Members Present	Members Absent	Others Present
<p>South Dakota Department of Health Beth Dokken, Division Director Family & Community Health Linda Ahrendt, Administrator, Office of Child & Family Services Jennifer Folliard, Assistant Administrator, Office of Child & Family Services Lauren Pierce, Newborn Screening Coordinator Bernadette Boes, Newborn Screening Coordinator Sarah Aker, South Dakota Medicaid</p> <p>Sanford Health Sanford Children’s Hospital NBS Medical Consultant Genetics/Metabolic Dr. Laura Davis-Keppen Dr. Isum Ward</p> <p>Indian Health Services Dr Carmen Ruiz, Pine Ridge, Pediatric Clinic</p> <p>Avera Health Alyssa Christensen, OB Nurse Manager</p>	<p>South Dakota Department of Health Dr Mary Carpenter, Medical Director</p> <p>Monument Health Kelli Sorenson, Lab Manager Nan Fitzgerald, NICU Manager</p> <p>Parent & Family Representative Jackie Vander Woude, Avera NICU Family Support</p>	<p>State Hygienic Laboratory at University of Iowa Stanton Berberich, Program Manager Travis Henry, Assistant Research Scientist Tate Kappell, Clinical Lab Supervisor Carol Johnson, Newborn Screening Follow up Program Coordinator</p> <p>SD EHDI Coordinator Shelby Hintze Jepperson, USD</p> <p>South Dakota Hands and Voices Jill Scott</p> <p>Other Public Miranda McAuliffe, ALD Alliance</p>

Agenda Item	Discussion
Welcome and Introductions	<ul style="list-style-type: none"> • Beth Dokken began the meeting at 10:00 am. Meeting took place in person & zoom • Introductions from SD DOH, committee members, and presenters.
Purpose of the Committee and Role of the Committee Members	<ul style="list-style-type: none"> • Jennifer Folliard provided overview of the purpose of the Newborn Screening Advisory Committee (NBSAC). The committee is intended to be advisory in nature meant to advise the South Dakota Newborn Screening (SD NBS) Program. • Future meeting topic: Development of Purpose Statement for South Dakota to help focus our efforts and communicate with the public. • Bernadette Boes, RN newest member of the SD newborn screening program was introduced. Her background was provided and is currently working with Lauren Pierce current coordinator in learning the role as Lauren is transitioning. Lauren will be leaving the role in August 2022 to return to further her education.
South Dakota Newborn Screening Program	<ul style="list-style-type: none"> • Bernadette Boes provided a presentation on the overview SD newborn screening bloodspot program, and the purpose and responsibilities of the state of South Dakota. The daily duties of the coordinator and follow up nurse were discussed. Numerical data reviewed number screened, follow up cases and positive screens. • Lauren Pierce reviewed Sharina's follow up program with a case study.
Spinal Muscular Atrophy (SMA) Screening Update	<ul style="list-style-type: none"> • SMA Screening began 9/1/21 no positives as of 4/19/21- 7561 babies have been screened • Average prevalence US 1:11,000 • Iowa 1:6,988, ND 1:21,500 • Travis Henry explained that the lab assay is very specific to an exon 7 deletion within SMN1. (95% of affected individuals) • He explained that deletion in the Hutterite population encompasses the same region as SMN1 exon 7.
Laboratory & Initial Follow-Up Services RFP Process	<ul style="list-style-type: none"> • Lauren Pierce reviewed the RFP process • October 2021: Released RFP • Pompe Disease Screening request included in RFP • December 2021: Proposals due 2 proposals received • January – February 2022: Evaluation process • Diverse evaluation team conducted in-depth evaluation • March 2022: Selected successful proposal: State Hygienic Laboratory at the University of Iowa • Pompe Disease Screening – Next steps: • Summer 2022 - Attend legislative Interim Rules Review Committee to prepare for Pompe disease screening

Worried that families will not understand importance of follow-up.

- Dr. Davis-Keppen said they have seen this with PKU. Families not following treatment until condition gets dangerous.
- Do we know why parents are hesitant to do follow-up treatment/care?
 - Cost
 - Follow-up appointment no-shows despite multiple attempts
 - At what point and in what situation should CPS become involved?
- Sarah Aker: Would be helpful to have added information about efficacy and outcomes of treatments for disorders Dr. Ward and Dr. Davis -Keppen covered. We will take that back and consider that as we develop agenda for future meetings.
- Beth Dokken: Does SD Medicaid cover treatment for any of these new conditions (MPS I, MPS II, X-ALD)? Another important consideration.
 - Dr. Davis- Keppen . These are similar to other things that are already on the newborn screening panel.
 - Children have been covered by Medicaid for these treatments in the past.
 - Cost associated with being involved in a clinical trial are generally covered by Medicaid but if drug is not approved by FDA, Medicaid will not cover it.
 - Not sure if gene therapy is covered by Medicaid. Not essential for a cure.
 - Beth recap: Medicaid does typically cover the therapy. Medicaid doesn't typically cover enzyme therapy unless FDA approved. Dr. Davis-Keppen indicated that most therapies have been FDA approved. There may be one drug for X-ALD that has not been approved. Something to continue to discuss.
 - Miranda (chat): ALD gene therapy review coming from FDA soon
- Dr. Davis-Keppen: Newborn hearing screening. Many children are identified with hearing loss late and it impacts language. Follow-up is not done with hearing in SD. Ensure that parents get appropriate education and that children get additional follow-up care. At least 1/3 are not getting diagnosed until later. She was advised DOH works with USD through the HRSA grant to improve testing.

<p>Public Comment Opportunity</p>	<ul style="list-style-type: none"> • Miranda Mc Auliffe , ALD Alliance: Mother of a son who diagnosed with X-ALD in NY 6 years ago. She is a partner with Elisa Sieger at ALD Alliance who advocate for ALD to be added to state newborn screening programs. Elisa’s son was diagnosed with ALD after lengthy diagnostic tests for months and has since passed away. Elisa turned her grief into advocacy and passion when she learned that there was a NBS for ALD. She started the ALD Alliance. Miranda joined the group when her son received this difficult diagnosis. Miranda learned of the diagnosis through newborn screening thanks to Elisa efforts in her state. Elisa’s son is happy and healthy 6-year-old boy. Many other of her own family members have been diagnosed and/or are carriers. They have been able to make educated decisions about growing families. ALD Alliance (created by Elisa) created a parent guide for ALD. Roadmap for families new to the diagnosis. Started a free care-package for ALD families. Running an ALD NBS consortium. She provided the following web sites • https://investor.bluebirdbio.com/news-releases/news-release-details/bluebird-provides-update-fda-review-timelines-betibeglogene • https://www.aldalliance.org/family-support.html • Beth Dokken: We will keep everyone updated via email with Pompe testing and moving through the administrative rule process.
<p>Discuss Next Meeting</p>	<p>Next SD NBSAC meeting – Our plan for these meetings going forward: We will continue hybrid model. Will meet annually in the spring but will put together the meeting earlier if needed</p>