



**SOUTH DAKOTA DEPARTMENT OF HEALTH**  
Every South Dakotan Healthy and Strong

**South Dakota Newborn Screening Advisory Committee**  
**Minutes**  
**April 7<sup>th</sup>, 2021**  
**2:00 pm to 4:00 pm**  
**Zoom call**

<b>Members Present</b>	<b>Members Absent</b>	<b>Others Present</b>
<p>Dr. Mary Carpenter Dr. Laura Davis-Keppen Dr. Isum Ward Kelli Sorenson Bill Snyder</p>	<p>Alyssa Christensen Nan Fitzgerald Dr. Carmen Ruiz</p>	<p>Beth Dokken – SD DOH Linda Arendt – SD DOH Jennifer Folliard – SD DOH Lauren Pierce – SD DOH Stanton Berberich – Iowa State Hygienic Laboratory (SHL) Travis Henry – Iowa SHL Tate Kappell – Iowa SHL Carol Johnson – Iowa Newborn Screening Program</p> <p>Ryan Colburn – public observer, oral comment Carolyn and Rory Philstrom – public observer, oral comment Margaret and Christopher Haberman – public observer, oral comment Donna Sachau – public observer, written and oral comments Maynard Friesz – CureSMA, oral comment</p> <p>Dee George – Novartis, public observer Jill Scott – South Dakota Hands &amp; Voices public observer Sandy Fawbush – HealthTech Solutions, public observer Shelby Hintze Jepperson – University of South Dakota, public observer Dan Kelly – public observer Joseph Hrdlicka – Genetech, public observer Amy Burke – ND NBS Program, observer</p>

Agenda Item	Discussion
<b>Welcome and Introductions</b>	<ul style="list-style-type: none"> <li>Beth Dokken began the meeting at 2:00 pm</li> <li>Introductions from SD DOH, committee members, and presenters.</li> </ul>
<b>Purpose of the Committee and Role of the Committee Members</b>	<ul style="list-style-type: none"> <li>Beth 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.</li> <li>Provided overview of process for adding new disorders to our Newborn Screening panel.</li> <li>Focus of meeting today: Discuss both Spinal Muscular Atrophy (SMA) and Pompe disease and make informed decision about how to proceed with each disorder.</li> </ul>
<b>Overview – South Dakota Newborn Screening System</b>	<p>Lauren Pierce provided a presentation on the overview and background of Newborn Screening Programs and South Dakota’s program. Main topics discussed include: the history of newborn screening in the US, how and when screening is done, SD NBS program statistics, the Recommended Uniform Screening Panel (RUSP), criteria for adding new disorders to the RUSP, the SD NBS panel of disorders, and the role of NBS Advisory Committees.</p>
<b>Spinal Muscular Atrophy (SMA) Overview and Magnitude of Benefit</b>	<p>Dr. Laura Davis-Keppen presented on SMA newborn screening in South Dakota. Topics of discussion: Experience and history with the SD NBS Program, history and purpose of the RUSP, history of SD NBS panel of disorders, support for adding RUSP conditions to the SD panel, overview of SMA including importance of early diagnosis via NBS and treatment, and experience with SMA in SD.</p> <ul style="list-style-type: none"> <li>Travis Henry asked question related to the increased frequency of SMA among the Hutterite population. Explained that the lab assay is very specific to an exon 7 deletion within SMN1. Asked if that deletion in the Hutterite population encompasses the same region as SMN1 exon 7. Would impact whether or not the lab can detect SMA among this population. Travis to investigate this further.</li> </ul>
<b>Insights from Iowa Newborn Screening Program</b>	<p>Carol Johnson shared the Iowa NBS Program’s experience with the SMA case they have identified in their SMA screening pilot in Iowa. Expressed importance of preparing the NBS system for the complexities of SMA screening, including the identification of an older sibling(s) with SMA.</p>
<b>Pompe Disease Overview and Magnitude of Benefit</b>	<p>Dr. Isum Ward presented on Pompe Disease and support for adding Pompe to SD’s NBS panel of disorders. Discussed treatment for Pompe, and importance of early detection and treatment.</p> <ul style="list-style-type: none"> <li>Dr. Davis-Keppen shared experience with treating Pompe disease and expressed the importance of early diagnosis and treatment and stated that outcomes are better with early treatment.</li> <li>Ryan Colburn provided additional perspective on treatment for Pompe disease. Dr. Ward expressed understanding and obligation to make clear that they have thought through possible barriers to treatment.</li> </ul>

<p><b>Implementation, Feasibility, Readiness, and Cost</b></p>	<p>Tate Kappell with the Iowa State Hygienic Laboratory (SHL), which is South Dakota’s contracted laboratory that conducts its newborn screening tests, presented on the assay used by the Iowa SHL for SMA screening (real time PCR) and provided a status update on their SMA screening pilot. Shared that they have found one presumptive positive SMA case since the beginning of the pilot in July 1, 2020. Plan to formally add SMA to Iowa panel and begin routine testing September 1, 2021. Shared feasibility of SD SMA screening and considerations to adding new disorders to a state panel. Travis stated that the Iowa SHL does not currently have the needed infrastructure to screen for Pompe disease and explained that building this infrastructure would require direction from the programs they service prior to adding those expenses. The anticipated timeline for adding Pompe screening would be approximately 36 months.</p> <ul style="list-style-type: none"> <li>• Discussion on the positive and challenging aspects of using multiple labs to screen for Pompe disease and that SD would need to conduct follow up research and coordinate logistics in order to add Pompe to our newborn screening panel.</li> </ul>
<p><b>Public Comment Opportunity</b></p>	<ul style="list-style-type: none"> <li>• Beth Dokken summarized written public comments. Received three written comments in support of adding SMA to newborn screening panel and one written comment in support of adding Pompe to panel.</li> <li>• Pastor Rory Philstrom, MN – Shared experience with son who has SMA that was detected on his newborn screen in MN and expressed support for adding SMA to SD’s NBS panel.</li> <li>• Margaret and Christopher Haberman, SD – Shared experience with son who was diagnosed with late-onset Pompe disease at age 3 and positive experience with treatment. Expressed support for adding both SMA and Pompe disease to SD’s NBS panel.</li> <li>• Maynard Friesz, CureSMA – Provided overview of SMA, importance of early diagnosis and treatment of SMA, and encouraged advisory committee members to recommend the addition of SMA to SD’s NBS panel.</li> <li>• Ryan Colburn – Shared personal story with Pompe disease and support for the addition of SMA, Pompe disease, and all RUSP conditions to the SD NBS panel.</li> <li>• Donna Sachau – Shared experience with granddaughter who has SMA and expressed support for adding SMA to SD’s NBS panel.</li> </ul>
<p><b>Committee Discussion and Recommendations</b></p>	<ul style="list-style-type: none"> <li>• Discussion about possible concerns with SMA screening assay as it relates to Hutterite population in South Dakota. Will need to investigate further.</li> <li>• Discussed assay for Pompe disease screening.</li> <li>• Dr. Davis-Keppen shared additional history of adding new disorders to the SD NBS panel and possibility of screening for Pompe with different lab if SD would like to before Iowa lab has needed infrastructure.</li> <li>• Bill Snyder confirmed that Medicaid covers SMA treatment within the criteria published on their website.</li> </ul>

	<ul style="list-style-type: none"> <li>• SMA Recommendation: Dr. Davis-Keppen recommended the addition of SMA to SD’s NBS panel. Dr. Ward seconded recommendation. All committee members recommended the addition of SMA to SD’s panel.</li> <li>• Pompe disease recommendation: Dr. Davis-Keppen recommended the addition of Pompe disease to SD’s NBS panel with the acknowledgement that there will be additional logistics to explore. Dr. Ward seconded recommendation. All committee members recommended the addition of Pompe disease to SD’s panel.</li> <li>• Beth shared that DOH will have further internal discussion and make a final decision for both disorders. Information and next steps regarding how SD will proceed will be shared with committee members and stakeholders as it becomes available.</li> </ul>
<b>Discuss Next Meeting</b>	Next SD NBSAC meeting – Fall of 2021 (in-person, pending COVID-19 precautions).
<b>Adjournment</b>	Meeting adjourned at 4:04 pm.