Newborn Screening Advisory Committee Meeting
Tuesday, October 28, 2025, from 8:30 a.m. to 12:00 p.m.
Nebraska Children's Home Society – Omaha

Committee Members	Specialty	Present	Absent
Peter Abasolo, MD	Pathologist	X	
Jill Allen, RN	Nebraska Hospital Association Rep.	X	
Khalid Awad, MD	Chair, Neonatologist	X	
Jill Beck, MD, MS	UNMC Pediatric Hematology/Oncology	X	
Angela Brennan, MD	Family Physician	X	
Amy Brower, PhD	Adjunct Professor of Genetics at Creighton University and Med Center	X	
Ria Fyffe-Freil, PhD	Clinical Chemist in Pathology	X	
Zoe González-García, MD	Pediatric Endocrinologist	X	
Jessica Hansen, MSN, FNP-C	APRN	X	
Kathryn Heldt, RD	Dietitian/Metabolic Specialty	X	
Isabella Herman, MD	Pediatric Neurogenetics	X	
Claire Jackson, MD	Hematology		X
Alyssa Keller, MS, CGC	Certified Genetic Counselor	X	
Tiffany Moore, PhD, RN	Parent Advocate	X	
Hana Niebur, MD	Vice-Chair, Pediatric Immunology	X	
Elizabeth Null, MD, FAAP, FACMG	Department of Pediatrics, Division of Inherited Metabolic Diseases, UNMC	X	
Erin Pierce, MD	General Pediatrics		X
Robert Rauner	X-ALD Advocate	X	
William Rizzo, MD	Pediatric Genetic/Metabolic	X	
Jill Skrabal, PhD, RD	Dietitian/Metabolic	X	
Heather Thomas, MD	CF Clinic Director	X	
Jennifer Wallin	Parent Advocate		X
Emeritus Members			
James Harper, MD	Pediatric Hematologist	X	
Richard Lutz, MD	Pediatric Genetic/Endo/Metabolic	X	
Deborah Perry, MD	Pathologist		X
Laboratory			
PJ Borandi	Operations Director Revvity Omics	X	
Meredith Patik	Revvity NBS Lab	X	
Staff	**Title**		
Erin Bryant, RN, BSN	Nurse Navigator, Hematology Sickle Cell Disease & Infant Hemoglobinopathies	X	
Jillian Chance, DNP, RN, APRN	NBS Program Manager	X	
Michaela Howard, RD	NBS Inherited Diseases Clinical Specialist	X	
Yousif Ibrahim	NBS Inherited Diseases Clinical Specialist	X	
Tyra Mills, MPH	Bioinformatics Fellow		X
Derek Ross	Birth Defects Registry Health Data Coordinator	X	
Krizia Sanoy	Newborn Screen Systems Coordinator	X	

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Tim Tesmer, MD	Nebraska DHHS CMO	**X**	
Sarah Ward, MPH	NBS Inherited Diseases Clinical Specialist	**X**	
Guests & Speakers			
Craig Baker, MD	Clinical and Biochemical Geneticist	**X**	
George Mr. De La Cerda, PharmD	Orchard Therapeutics Guest Speaker	**X**	
Catherine Harbert, MS, CHG	Pediatric Genetic Counselor	**X**	
Susheela Ms. Jayaraman, MS, MA, CGC	Orchard Therapeutics Guest Speaker	**X**	
Missy Moody	Neuro Genetics Case Manager	**X**	
Victoria Shum, MD	Fellow	**X**	
Lois Starr, MD	Chief UNMC Division of Genetics	**X**	
Sarah Sutter, MD	Pediatric Resident	**X**	
Abby Wolfe, MD	Pediatric Resident	**X**	
Rhonda Wright, MD	Pediatric Neurologist	**X**	

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Open Meeting Act Announcement

Dr. Khalid Awad, the chair, called the October 28, 2025, quarterly meeting for the Nebraska Newborn Screening Advisory Committee to order at 8:36 a.m. He announced that, under the Nebraska Statute (Open Meeting Act), a notice of this public meeting is posted on the State of Nebraska Public Meeting Calendar, and a copy of the Open Meeting Act and the meeting agenda are posted on the wall by the entrance to the meeting space.

Introduction

Dr. Khalid Awad asked the guests, personnel, advisory committee members, and new advisory committee members to introduce themselves. Craig Baker, MD (Clinical and Biochemical Geneticist), George Mr. De La Cerda, PharmD (Orchard Therapeutics), Catherine Harbert, MS, CHG (Pediatric Genetic Counselor), Susheela Ms. Jayaraman, MS, MA, CGC (Orchard Therapeutics), Missy Moody (Neuro Genetics Case Manager), Victoria Shum, MD (Fellow), Lois Starr, MD (Chief UNMC Division Genetics), Sarah Sutter, MD (Pediatric Resident), Abby Wolfe (Pediatric Resident), and Rhonda Wright, MD (Pediatric Neurologist) all attended as guests.

Review/Approval of Minutes from the April 2025 Meeting

The approval of the July 29, 2025, meeting minutes was brought up by Dr. Khalid Awad. The motion to approve was made by Dr. Zoe González-García, seconded by Dr. Angela Brennan, and passed unanimously by all committee members.

Laboratory Update

Revvity Omics provided an update regarding a new platform currently under consideration by the testing laboratory. Revvity Omics noted that implementation of this platform will necessitate adjustments to laboratory cut-offs, contingent upon validation results. Revvity Omics indicated that additional information regarding the platform's impact on existing systems will be disseminated as it becomes available. The company also reported ongoing expansion of its genomics offerings. It announced the forthcoming launch of Metachromatic Leukodystrophy (MLD) testing, with implementation by one state client that commenced in January this year.

Krizia Sanoy, Coordinator for the Newborn Screening Program Systems and Metabolic Foods, provided an update concerning UPS shipping delays. During the third quarter of 2025, a total of 137 specimens experienced preventable delays, exclusive of 105 delays attributable to unavoidable circumstances. In July, 88 specimens were delayed due to avoidable delays; by October, this number had decreased to 26. Ms. Sanoy indicated that this improvement is attributable to the implementation of corrective actions following the August 22 meeting with UPS. Nevertheless, the NBS program noted that delays have increased over the past two weeks due to missed pickups and sortation issues. Revvity is currently undertaking measures to mitigate further disruptions.

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Dr. Jillian Chance reported that the meeting with UPS held on August 22 yielded proactive outcomes aimed at addressing and preventing avoidable delays. She further noted that the program coordinator has been diligently working with Revvity to secure the provision of pre-printed, prepaid shipping labels. These labels are intended to be kept in stock at participating hospitals, enabling continued specimen shipment in the event of ShipExec system downtime. This measure is designed to prevent delays by allowing the hospitals to dispatch specimens without interruption.
Dr. Chance emphasized that, while this may appear to be a simple solution, it represents a significant logistical and financial undertaking, as Revvity must cover the cost of all pre-printed labels in advance to be stored at hospital sites. Nevertheless, this initiative is expected to make a meaningful difference in maintaining timely shipments, particularly when printing issues arise. She further acknowledged the substantial collaborative efforts made over the past several months by UPS and Revvity, in coordination with the state's newborn screening program, to minimize delays and ensure that specimens are consistently collected and shipped on schedule.

Legislation, Regulation, and Policy

NBS program staff informed the State's Newborn Screening Advisory Committee that the Secretary's Advisory Committee remains inactive and that, at this time, the Health Resources and Services Administration (HRSA) has no official plans for its reinstatement. Program personnel clarified that the Recommended Uniform Screening Panel (RUSP) remains the federally recognized standard. Program staff explained that the Recommended Screening Panel, which is codified in federal law, had previously been moved under a statutory framework. Following the reauthorization of the Newborn Screening Saves Lives Act in 2019, the committee's status transitioned to a voluntary basis, allowing for its subsequent dissolution. However, the RUSP remains in effect. They further noted that recent actions concerning the two most recent conditions under review —Duchenne Muscular Dystrophy and Metachromatic Leukodystrophy (MLD) — have been published in the Federal Register. Public input has been solicited regarding these conditions. Additionally, program staff reported that the American College of Medical Genetics and Genomics (ACMG) has initiated efforts to establish an alternative or supplementary body to the Secretary's Advisory Committee on Genetics, Health, and Society.

For State updates, the Program noted that they have not received notice of any pending or proposed legislation directly affecting the newborn screening program. The NBS program further indicated that an official hearing is anticipated concerning updates to the inclusion of Mucopolysaccharidosis Type II (MPS II) and Critical Congenital Heart Disease (CCHD) regulations. These updates will require hospitals to submit all CCHD-related data and to implement the updated American Academy of Pediatrics (AAP) Algorithm for screening and reporting.

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Presentation on Metachromatic Leukodystrophy (MLD) by Orchard Therapeutics

On behalf of Orchard Therapeutics, Medical Science Liaison George De La Cerda presented on Metachromatic Leukodystrophy (MLD), a rare inherited lysosomal storage disorder that profoundly affects the nervous system. Joined by his colleague, Ms. Susheela Jayaraman, Mr. De La Cerda provided a comprehensive exploration of the disease's scientific foundations and the significant recent advances in its diagnosis, treatment, and screening initiatives.

Mr. De La Cerda began by describing MLD as a progressive, autosomal recessive condition caused by a deficiency of the enzyme arylsulfatase A (ARSA). The absence of functional ARSA leads to the accumulation of sulfatides within lysosomes, resulting in widespread demyelination throughout the central and peripheral nervous systems. Over time, this pathological process causes severe cognitive and motor deterioration, ultimately leading to early mortality. Though MLD is considered rare, occurring roughly one in every 100,000 live births, Mr. De La Cerda noted that the actual prevalence is likely higher. Specific populations, including Navajo and Israeli Arab communities, have shown elevated carrier frequencies, suggesting underdiagnosis in the general population.

MLD is classified primarily by the age at which symptoms appear. The late infantile form, the most severe, typically presents before the age of two. The early juvenile form emerges between two and seven years, while late-onset forms occur later in life. Earlier onset usually correlates with faster disease progression and more profound neurological impairment. Without newborn screening, diagnosis requires a combination of biochemical and genetic tests, including measurement of ARSA enzyme activity in leukocytes, assessment of urinary sulfatide levels, and confirmatory genetic sequencing to identify pathogenic variants. Although most infants appear normal at birth, they soon exhibit developmental regression, losing speech, motor function, and cognitive skills as the disease advances.

Turning to treatment, Mr. De La Cerda introduced Atidarsagene Autotemcel, marketed as Arsa-cel, the first FDA-approved gene therapy for MLD. Designed for children with late infantile or early juvenile forms, Arsa-cel uses the patient's own CD34+ hematopoietic stem cells, which are genetically modified to express a functional copy of the ARSA gene. The therapy process is intricate yet highly controlled. Eligible patients are referred to one of several certified U.S. treatment centers located in Minnesota, San Francisco, Houston, Atlanta, and Philadelphia, where their stem cells are collected via apheresis. These cells are then transported to a specialized manufacturing facility in Milan, Italy, where they undergo genetic modification and extensive quality control over a period of six to seven weeks. Once processed, the modified cells are shipped back to the treatment center, where the patient undergoes a myeloablative conditioning regimen before receiving the cell infusion. Patients remain under close medical supervision for several weeks following treatment.

Mr. De La Cerda reviewed the therapy's clinical development program, which included two formal clinical trials and an expanded-access cohort. In total, 39 patients received Arsa-cel and were compared to 49 untreated patients in a natural history cohort. The results were remarkable: treated patients demonstrated preserved motor function, improved survival, and retained

cognitive abilities. At the time of analysis, 100% of treated patients were alive, compared to approximately 40% in the untreated group. Many children who received Arsa-cel maintained the ability to communicate and learn, representing a profound improvement in quality of life. Safety findings aligned with expectations for myeloablative stem-cell therapy; most adverse events, such as transient neutropenia, were related to conditioning rather than the gene therapy itself. Crucially, there were no treatment-related deaths or instances of insertional oncogenesis.

Following Mr. De La Cerda's presentation, Susheela Jayaraman emphasized the critical importance of newborn screening for MLD. She explained that early detection is essential, as most children are currently diagnosed only after symptoms appear, often too late to qualify for gene therapy. Data from the United Kingdom revealed that many children referred for MLD evaluation were already too symptomatic for treatment, with several treated cases being younger siblings of previously diagnosed patients.

Ms. Jayaraman outlined the three-tier newborn screening algorithm, which was developed through international collaboration. The process begins with the measurement of sulfatides in dried blood spots, followed by testing for ARSA enzyme activity. It concludes with gene sequencing to confirm diagnosis and distinguish actual disease from benign pseudodeficiencies. Pilot studies have validated this approach. A landmark German study screened over 100,000 newborns, identifying three actual cases of MLD without any false positives. Two of these infants, both with early-onset forms, were treated with Arsa-cel and demonstrated positive outcomes. Similar pilot programs are now active globally, including in Norway and several U.S. states, such as Illinois, Minnesota, Pennsylvania, and New York, the latter of which recently implemented statewide MLD screening under a federal research initiative.

Ms. Jayaraman also highlighted the growing body of expert consensus guidelines from both U.S. and European panels that now inform the diagnosis, monitoring, and treatment of MLD. The U.S. Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) has completed a favorable evidence review supporting the inclusion of MLD in the federal Recommended Uniform Screening Panel (RUSP). However, final approval awaits further government action following the committee's disbandment.

In closing, Ms. Jayaraman underscored Orchard Therapeutics' continued leadership in advancing MLD research, screening, and patient access. Since 2020, the company has led the MLD Newborn Screening Alliance, a coalition of clinicians, researchers, and advocacy organizations dedicated to expanding early detection and treatment opportunities for MLD. The alliance alternates annual meetings between Europe and the United States, fostering collaboration, data sharing, and policy development across the global MLD community.

Together, George, Mr. De La Cerda, and Susheela Jayaraman conveyed a shared vision of a future in which early detection, equitable access, and scientific innovation transform the trajectory of MLD. Through the integration of gene therapy and newborn screening, they envision a world where children affected by this devastating disease are not only able to survive but to live whole, meaningful lives.

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Bioinformatics Fellow Update

For the Bioinformatics fellow update, Yousif Ibrahim, one of the follow-up clinical specialists, stood in for Tyra Mills, a fellow who had been leading several data visualization initiatives within the Nebraska Newborn Screen program. In Ms. Mills' absence, her colleague presented the findings and insights she had prepared, offering an overview of how geographic distance, provider referral patterns, and healthcare facility characteristics influence access to hematology care, particularly for patients affected by sickle cell disease and related conditions.

The presentation began with an introduction to a Power BI dashboard developed by Ms. Mills. The first visualization displayed referral states by distance from the hematology clinic. In this stacked column chart, red bars represent individuals who were not referred, while green bars represent those who were referred. The data revealed that approximately 70% of the study population lived within 25 miles of a hematology clinic. This figure is consistent across both pre- and post-intervention datasets. Within this group, forty-seven percent of participants were not referred, while twenty-two percent were referred. Among participants living between twenty-six and seventy-five miles away, 10% were not referred, and 11% were referred. Finally, among those living more than 76 miles from the clinic, 7% were not referred, and 3% were referred. These patterns suggested that distance alone was not the sole determinant of whether patients received a referral for specialized hematology care.

The second visualization examined whether referred patients ultimately attended their appointments. In this chart, the black segments represent individuals who were referred but not seen in clinics, while the green segments represent those who were both referred and seen. The analysis showed that twenty-two percent of participants living within twenty-five miles were referred, yet only twelve percent of those individuals actually attended a clinic visit. This finding suggests that factors other than distance, such as transportation challenges, scheduling barriers, or communication gaps, likely contributed to the discrepancies between referral and attendance rates.

A third visualization explored referral rates by the size of the birth facility. Using the same color-coding system, the analysis demonstrated no significant difference in referral rates across facilities of varying sizes. Regardless of the size of the birth facility, most patients were not referred more frequently than they were referred. This outcome suggested that systemic factors, rather than institutional size or capacity, may have a greater impact on referral decisions.

When the data was filtered to include only providers with medical degrees, and when non-metropolitan areas were isolated, further insights emerged. The filtered dataset focused on participants diagnosed with sickle cell disease or sickle cell trait. Red segments represented those not referred to or seen, black represented those referred to but not seen, and green represented those both referred to and seen. Out-of-state participants were included to maintain completeness, with bubble size corresponding to the number of cases in each region. Through the use of Power BI's mapping and visualization features, the research team was able to display these complex relationships in a clear and accessible format, enhancing understanding of the data's geographic and demographic dimensions.

The presentation also recognized the substantial contributions of Ms. Mills to the project. Although her fellowship had concluded and she had accepted a new position with the Virginia Newborn Screening Program, the Power BI dashboards she developed would continue to serve as valuable analytical tools for ongoing research. These dashboards were designed to facilitate continuous data collection, monitoring, and visualization, ensuring that the hematology program could make data-driven decisions to improve patient outcomes. Her work exemplified how digital tools can transform raw data into actionable insights, strengthening the link between research and clinical practice.

Following the presentation of findings, Dr. Awad raised an essential question regarding the low number of referrals for patients diagnosed through newborn screening with hemoglobinopathies. Dr. Harper provided clarification, noting that while patients with confirmed sickle cell disease are routinely referred to specialty care, those identified with sickle cell traits are often managed within their primary care home. This approach is designed to reduce unnecessary specialty visits for patients who are carriers rather than those who are symptomatic. However, Dr. Harper also acknowledged that inconsistencies in newborn screening reports, particularly when primary care providers are not correctly listed, can lead to lapses in communication and follow-up. As a result, some families may remain unaware of their child's carrier status or fail to receive appropriate counseling.

The discussion expanded to address broader challenges affecting patient access to care. In Nebraska and other regions of the Midwest, vast geographic distances and limited funding for outreach clinics have historically restricted access to specialized hematology services. Without adequate resources to sustain outreach programs, many rural patients face significant travel burdens. Despite these limitations, the team has sought creative solutions to bridge these gaps. Telemedicine initiatives and the introduction of a mobile clinical unit are among the strategies being implemented at the Children's Hospital. The mobile unit is expected to support school-based and community outreach, providing essential follow-up services such as transcranial Doppler ultrasounds for patients who cannot easily travel to clinic appointments. Community engagement was also emphasized as a crucial component of improving access and awareness. Collaboration with organizations like the Owens Foundation has played a vital role in expanding outreach to families affected by sickle cell disease and trait across state lines. A prime example of such efforts was the annual Sickle Cell Education Day hosted by the Children's Hospital. Now in its third year, the event brought together patients, families, healthcare providers, and community advocates for a day of education, support, and celebration. The event featured breakout sessions, informational booths, and activities for children, promoting both learning and community engagement. The Kroc Center's generous donation of space and resources contributed to the event's success, reflecting the strong partnerships that continue to drive these initiatives forward.

In conclusion, the Program's work highlights the power of data visualization and interdisciplinary collaboration in addressing healthcare disparities. By leveraging Power BI dashboards, promoting telehealth access, and engaging community partners, the Program has made significant progress toward ensuring equitable care for individuals with hematologic

conditions. Through Ms. Mills' foundational work and the continued dedication of the team, these efforts exemplify how data-informed decision-making can shape a more inclusive and responsive healthcare system.

Case Management System Update

Derek Ross, the Health Data Coordinator for the Nebraska Birth Defects Registry, introduced the Birth Defects Registry Dashboard, a project jointly developed over the past six months by himself and his colleague, Ms. Mills. He noted that while he contributed to the data and assisted in identifying meaningful variables and visualizations, the majority of the dashboard's design and functionality were primarily the result of Ms. Mills' efforts.

The presentation began with an overview of the project's background and context. The Nebraska Birth Defects Registry has not released data to the public since 2016 - a problem the dashboard aims to remedy. At that time, data were presented through an annual statistical report that included statewide information on births, deaths, congenital anomalies, marriages, divorces, and other vital statistics. In recent years, the Nebraska Department of Health and Human Services (DHHS) has transitioned from static reports to interactive dashboards for presenting public health information. These include dashboards for topics such as abortion statistics, tick surveillance, and other health indicators. The Birth Defects Dashboard is designed to follow this model, offering a modern and accessible format for data dissemination.

Mr. Ross emphasized that the dashboard remains a work in progress and clarified that all data displayed are provisional and subject to revision. An accompanying "About the Data" document is under development to provide additional transparency regarding data sources, methodologies, and definitions. The dashboard currently includes data spanning five years from 2019 through 2023. It focuses on the 47 major congenital anomalies as defined by the National Birth Defects Prevention Network (NBDPN), one of the nation's leading authorities on birth defect surveillance. The registry includes only defects reported among live births or fetal deaths of Nebraska resident mothers. While the registry accepts reports on out-of-state births and fetal deaths to Nebraska resident mothers, Mr. Ross explained that data collection from out-of-state sources was more comprehensive during the earlier years of the dataset, as recent limitations have reduced that capability.

Current suppression rules are applied to protect confidentiality. Counts between one and five are suppressed, and counts between six and ten are converted to percentages. For demonstration purposes, only aggregate data with sufficient case counts were displayed to avoid suppression-related complications. Only the first page of the dashboard, which contains the most general, least granular information, is tentatively being considered for future public release pending internal review and approval.

Mr. Ross provided a guided overview of the interface, focusing on the "Public" tab, which is designed to provide a level of detail comparable to the previously published annual reports. The dashboard enables users to explore birth defect counts by year and various demographic or clinical variables, including maternal age, maternal race, and defect category. Summary statistics

for the five-year period are displayed at the top of the dashboard for context. He clarified that visualized counts represent individual defect instances ("items") rather than unique cases, meaning a single child with multiple defects may contribute more than one item to the total.

The dashboard's interactive design allows users to filter and analyze data dynamically. Selecting a demographic category or variable automatically updates all corresponding charts and tables. For example, users can isolate data for female infants or mothers of a specific racial or age group to examine how these factors correlate with specific congenital anomalies. Slicers further enhance this interactivity, allowing users to refine results and identify particular patterns, such as cardiovascular defects among mothers of advanced maternal age, with just a few selections.

Mr. Ross highlighted the dashboard's potential applications for both researchers and public health professionals. Because the registry has not released public data in several years, data requests often require lengthy processing. By publishing a public-facing version of the dashboard, researchers could independently access the information they need, streamlining the data access process and reducing administrative workload.

He also discussed planned enhancements, including the addition of a mobile-friendly interface to improve accessibility and compliance with Americans with Disabilities Act (ADA) standards. The development team has drawn inspiration from the Tennessee Birth Defects Dashboard and other Nebraska DHHS dashboards to ensure consistency in design, suppression rules, and branding. However, the dashboard remains under development, and both internal and public release versions are still subject to further approval and refinement.

Mr. Ross provided a brief preview of the internal-use-only features, such as the "Risk Factors" tab. This internal tab visualizes relationships between birth outcomes – such as low birth weight or preterm birth – and factors like maternal or paternal age, access to care, and county of residence. He also described the "Geography" tab, designed to map the distribution of congenital anomalies across Nebraska by ZIP code and county, providing valuable spatial insights for surveillance and prevention planning.

Finally, Mr. Ross noted that Power BI's flexible framework allows for continued expansion and customization of the dashboard. Future updates may include additional analytical tabs and export functionality to support research requests and public records requests, thereby increasing the registry's efficiency and public health value.

Mr. Ross gave an update on the Program's implementation of the Natus Case Management System, a comprehensive and fully integrated digital platform designed to modernize the state's newborn screening data management processes. This system represents a significant advancement from the Program's longstanding reliance on manual data entry and Excel spreadsheets. By consolidating multiple functions into a single, secure environment, Natus is expected to improve data accuracy, streamline communication, and enhance the overall efficiency of public health program operations.

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Mr. Ross provided an overview of the Natus system, using screenshots obtained directly from the Natus demonstration environment. Although the demonstration does not yet reflect the Program's final customized version, it provides an accurate representation of the platform's basic functionality and design. All images utilized were based on scrambled data to ensure that no personally identifiable information was displayed. The Program emphasized that the system remains under development and that the current version does not represent the finalized product. Adjustments, refinements, and program-specific configurations will continue to be made before full implementation.

The transition to Natus aligns with the Program's long-term goal of modernizing data management across newborn screening programs. The current reliance on spreadsheets and manual tracking methods has long been a major limiting factor in the Program's ability to efficiently coordinate follow-up activities, manage case information, and communicate with providers. The Natus platform is being implemented to serve as a centralized, unified system for collecting, managing, and reporting newborn screening data. Its primary objectives include:

- Integrating information from multiple data sources, including vital records, laboratory results, and screening outcomes.
- Improving communication with healthcare providers and facilities through secure, direct channels within the system.
- Allowing external partners to access data relevant to their roles through customized user permissions.
- Automating key operational processes such as letter generation, record matching, and reporting, thereby reducing human error and administrative workload.

This system represents a shift toward a more efficient, transparent, and data-driven approach to managing newborn screening activities across the state.

Upon signing into Natus, users are greeted with a customizable dashboard designed to provide an at-a-glance overview of critical data. The dashboard highlights key indicators, such as unmatched laboratory results, pending transfers, unsatisfactory screenings, and pending correspondence. Each user's dashboard is tailored according to their assigned role, ensuring that information displayed is both relevant and actionable.

At the core of Natus is the Case Management System, which serves as the foundation of the platform. This feature enables users to create, manage, and track individual cases, linking each to relevant documents, providers, and facilities. The system also allows the assignment of specific tasks through an Action Manager, which tracks pending and completed actions on a case-by-case basis.

Natus includes multiple program-specific modules designed to address the unique needs of newborn screening programs. The Dried Blood Spot (DBS) module, for example, displays test results in a clear and organized format, with unsatisfactory results visually flagged. Similarly, the Hearing Screening and Critical Congenital Heart Disease (CCHD) modules provide accessible

and standardized data displays. A Birth Defects module is also under development and will be the first of its kind implemented through the Natus platform for the Program.

A defining feature of Natus is its ability to integrate with external systems through HL7 data transmission, a secure electronic data exchange standard widely used in healthcare. This functionality will enable newborn screening results to flow automatically from laboratories into Natus, removing the need for manual data entry.

The same level of integration extends to hearing screening and CCHD equipment. Facilities equipped with compatible devices will be able to connect directly to the system for automatic data upload. For those without HL7 capabilities, Natus will allow quick and secure data file imports, significantly reducing manual effort and potential delays. This level of automation is expected to enhance both efficiency and accuracy, thereby minimizing the administrative burden on hospital and clinic staff while ensuring timely access to critical data.

Natus is built around a role-based access control framework. Each user, whether a clinician, facility representative, or program staff member, will have defined permissions to determine what information can be viewed, edited, or managed.

Clinicians will have the ability to access newborn screening results, confirm diagnoses, and close cases directly within the system. In instances of abnormal screening results, the system can automatically generate and route referrals, notifications, and case updates. This process will replace the manual workflows currently used, such as faxing or emailing letters individually. While traditional fax communication may remain an option during the transition period, the long-term goal is to handle all correspondence and case management activities securely and electronically within Natus.

The Natus system offers a range of benefits that extend beyond convenience and modernization. It introduces an infrastructure that is responsive, intuitive, and scalable, capable of supporting the complex and evolving needs of public health programs.

Once fully implemented, Natus will:

- Eliminate redundant manual data entry and reduce administrative workload.
- Enhance communication between the Program, healthcare providers, and birthing facilities.
- Enable faster access to newborn screening results and follow-up data.
- Support accurate and timely reporting for state and federal requirements.
- Improve data integrity through automated validation and secure information exchange.

For clinicians, Natus will serve as a reliable resource for accessing newborn screening results, hearing screening outcomes, and CCHD findings for patients under their care. This capability is expected to simplify clinical workflows, improve continuity of care, and support timely medical interventions when needed.

The introduction of Natus represents a significant milestone in the Program's digital transformation efforts. The system's flexibility allows for ongoing development, expansion, and refinement as funding and time permit. Additional modules and functionalities can be added post-launch, ensuring that Natus continues to evolve in alignment with program needs and technological advancements.

Ultimately, Natus will replace outdated spreadsheet-based systems with a comprehensive, secure, and automated case management platform. It will facilitate more effective collaboration among facilities, providers, and public health programs while positioning the Program to better meet the challenges of data management in the modern era.

The Program acknowledges the extensive effort and collaboration required to bring this project to fruition. It expresses its appreciation to all program staff, healthcare providers, and partners who have contributed to the successful implementation of this system thus far. The transition to Natus signifies not only a technical upgrade but also a meaningful step toward efficiency, accuracy, and progress in newborn screening services statewide.

Request To Place Information on The Department Website Regarding How to Add a Condition. Discussion On Creating a Nomination Form for Advocates to Fill Out

During the recent committee meeting, Dr. Chance reported that she had been contacted by a parent advocate at the Association of Public Health Laboratories (APHL) Newborn Screening Symposium. The advocate requested that Dr. Chance share with the committee a one-page proposal recommending that all states establish a standardized, publicly accessible nomination process for adding disorders to their state newborn screening panels.

According to the advocate, many state newborn screening programs currently lack clear, publicly available information outlining how individuals or advocacy groups may propose the inclusion of new conditions. She intends to ensure that members of the public beyond established advocacy organizations can easily identify the appropriate procedure for submitting such recommendations. The advocate emphasized the importance of transparency and accessibility, suggesting that each state maintain a publicly available form or description of its nomination process on the website for its newborn screening program.

The committee discussed this proposal at length. Members generally expressed support for the concept, recognizing that the public should have access to clear guidance on how to nominate conditions for consideration. However, several members raised practical concerns, including the potential administrative burden that could arise if the Program received a large volume of submissions, particularly from organized advocacy groups. The committee acknowledged the need to balance public accessibility with the Program's capacity to review and evaluate nominations effectively.

It was noted that while the current process allows individuals or organizations to bring recommendations directly to the committee, there is no formalized or standardized nomination form in place. The committee discussed the possibility of creating such a form and developing

accompanying criteria for evaluating proposed additions. These criteria include considerations such as the availability of effective treatments, the clinical validity and utility of available screening tests, and the overall public health impact of adding the condition to the panel.

Several members emphasized the importance of defining and publicly communicating the evidence standards used to evaluate potential additions to the system. They noted that clear criteria, such as requirements for demonstrated clinical benefit, treatment feasibility, and cost-effectiveness, would help manage expectations and prevent inappropriate or non-evidence-based nominations. Providing this information alongside the nomination form could also serve an educational purpose, helping the public understand the rigorous evaluation process that underlies the composition of the newborn screening panel.

Members also discussed the potential for a subcommittee or working group to examine the proposal in greater detail. This subcommittee would be tasked with drafting a standardized nomination form, reviewing existing evaluation criteria, and recommending methods for public dissemination of the information. The group would also seek input from the Department of Health and Human Services and other relevant stakeholders to ensure consistency with existing state and federal practices.

The committee agreed that this matter merits further exploration. It was proposed that a small working group be convened to review the existing framework for evaluating proposed diseases, assess how it could be made publicly available, and prepare recommendations for the committee's consideration at its next scheduled meeting in January.

In summary, the committee recognized the value of enhancing public transparency and accessibility in the newborn screening nomination process. Members agreed that developing a standardized, publicly available submission mechanism could improve community engagement while maintaining the scientific rigor and procedural integrity of the newborn screening program.

APHL Everyday Life Saver Award

During the annual Association of Public Health Laboratories (APHL) Newborn Screening Symposium, held to recognize outstanding contributions to public health and newborn screening, eight awards were presented to honor exemplary professionals and advocates in the field. Initially, six awards were scheduled to be distributed; however, due to the exceptional quality of the nominees, two additional awards were granted. Among this year's distinguished recipients was Dr. Jill Skrabal, who received the award.

The Everyday Lifesaver Award recognizes individuals working in newborn screening or family advocacy who have made significant contributions to advancing the discipline. This award intends to honor professionals whose ongoing efforts contribute to the morale and efficiency of newborn screening programs, as well as those who promote the growth, knowledge sharing, and collaborative spirit that underpin the newborn screening system. Dr. Skrabal's recognition was strongly supported by testimonials from colleagues and families whose lives have been positively impacted by her professional excellence and compassionate care. One such

testimonial, provided by a parent whose child was diagnosed through newborn screening, reads as follows:

"My son, Bentley, was diagnosed six years ago with Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) through newborn screening. From the first day, Dr. Skrabal guided our family with compassion and clarity. Within hours of receiving Bentley's positive screening result, she translated complex medical information into clear, understandable language and developed an emergency care plan we could confidently follow. Since that day, she has remained our steadfast ally. Dr. Skrabal's unique combination of clinical skills, empathy, and dedication has transformed what could have been an overwhelming diagnosis into a manageable part of our lives. No family could ask for a better advocate or champion. Beyond her professional duties, Dr. Skrabal has gone above and beyond to support our family. She has taken after-hours calls to discuss urgent concerns regarding our son. She has even provided personal guidance to my husband following his diagnosis with diabetes, assisting him in developing a sustainable and effective diet plan. Over the years, our relationship has grown beyond that of clinicians and family. Dr. Skrabal attends our annual "Run for Thomas and Bentley," an event dedicated to raising awareness for MCAD and other Fatty Acid Oxidation Disorders (FAODs). She has continually supported our efforts to educate the community and to honor the memory of Thomas, who passed away due to MCAD, while celebrating Bentley's continued strength and health. Bentley greets Dr. Skrabal with great joy whenever they meet, whether at an appointment or by chance in the community. She continues to support us through regular communication and virtual check-ins, always offering encouragement and understanding. We are deeply grateful for her commitment and care. She is more than deserving of this recognition."

In addition to her clinical excellence, Dr. Skrabal has made significant contributions to the culture of recognition within the newborn screening community. Dr. Chance created a professional keepsake containing photographs, award letters, and commemorative materials for Dr. Skrabal, ensuring the preservation of these meaningful achievements.

Upon receiving her award, Dr. Skrabal expressed her gratitude, stating:
"We were recently discussing the importance of healthcare's dedicated workers, and I believe I have been relabeled as a metabolic nutrition bounty hunter. I truly love what I do and am deeply honored by this recognition. It is the patients and their families who make this work so meaningful, and I cannot imagine doing anything else."

Dr. Skrabal's exemplary leadership, professionalism, and compassion reflect the highest standards of the newborn screening community. Her recognition is a testament to her enduring commitment to patient care, public health, and the advancement of newborn screening services.

Other Business

No discussion was held on this agenda item, as there were no matters requiring debate.

Newborn Screening Advisory Committee Meeting
Tuesday, October 28, 2025, from 8:30 a.m. to 12:00 p.m.
Nebraska Children's Home Society – Omaha

Data

A closed session of members and staff was held for review of data. Information regarding Nebraska births, including births screened, unscreened births, out-of-hospital births, out-of-hospital births screened, unsatisfactory specimens, confirmed positive cases, confirmed negative cases, repeated typical cases, and pending cases, was provided.

Adjourned

Dr. Khalid Awad adjourned the meeting at 11:14 a.m.

Respectfully Submitted:

Jillian Chance
Michaela Howard
Yousif Ibrahim
Tyra Mills
Derek Ross
Krizia Sanoy
Sarah Ward