Rare Disease Subcommittee

Meeting Minutes

June 9, 2023
11:00 a.m.

Location: Microsoft Teams

Table 1. Rare Disease Subcommittee member attendance at the Friday, June 9, 2023, meeting

MEMBER NAME	IN ATTENDANCE
Khrystal Davis, J.D. (Subcommittee Chair)	Yes
Beryl Pamela "Pam" Andrews	Yes
Melissa Frei-Jones, M.D. (Ex-officio)	No
Charleta Guillory, M.D., M.P.H.	Yes
Barbra Novak, Ph.D., C.C.C.-A.	No
Michael Speer, M.D. (Ex-officio)	Yes

Table 2. Texas Department of State Health Services (DSHS) staff attendance at the Friday, June 9, 2023, meeting

ATTENDEE NAME	IN ATTENDANCE
Laura Arellano, Unit Coordinator, DSHS Newborn Screening (NBS) Unit	No
Karen Hess, Director, DSHS NBS Unit	No
Patricia Hunt, Newborn Metabolic Screening Group Manager, DSHS Laboratory	Yes
Aimee Millangue, Advisory Committee Liaison, DSHS NBS Unit	Yes
Rachel Lee, Ph.D., Medical Screening Unit Director and NBS Technical Advisor, DSHS Laboratory	Yes
Susan Tanksley, Ph.D., Deputy Laboratory Director, DSHS Laboratory	Yes

Previous Subcommittee Business

The Rare Disease Subcommittee (subcommittee) last met by Microsoft Teams on January 6, 2023. The subcommittee reported on their August 10, 2022 and January 6, 2023 to the full Newborn Screening Advisory Committee (NBSAC) at the March 24, 2023 NBSAC meeting. In July 2021, Ms. Lisa Otto, a parent of a child with Noonan Syndrome, provided public comment regarding the condition at a NBSAC meeting. Ms. Otto was invited to present to the subcommittee at their June 9, 2023 meeting.

Subcommittee Meeting Notes

Ms. Khrystal Davis, Subcommittee Chair, called the meeting to order at 11:06 a.m. Aimee Millangue, Advisory Committee Coordinator and Ombudsman, DSHS NBS Unit, recorded and transcribed the meeting through Microsoft Teams. Ms. Davis greeted everyone, and everyone in attendance provided brief introductions. Due to technical difficulties, Ms. Davis turned the floor over to Ms. Pam Andrews, subcommittee member, to preside over the meeting. Ms. Andrews then turned the floor over to Ms. Lisa Otto to present to the subcommittee. Ms. Otto referenced the PowerPoint, *An Overview of Noonan Syndrome* and the video *What are the RASopathies (updated).* After her presentation, Dr. Susan Tanksley and Dr. Rachel Lee provided a legislative update.

Noonan Syndrome

Highlights of the presentation included:

- Ms. Otto has a personal mission to raise awareness, make changes, and advocate for those who have been diagnosed with Noonan Syndrome in hop to improve their lives as well as caregiver support.
- The youngest of her three children was diagnosed in 2013 with Noonan Syndrome.
- She also has 30 years of experience as a pediatric occupational therapist and has worked in many areas of the hospital but is most passionate about babies and enjoys her days in the Neonatal Intensive Care Unit (NICU).
- The purpose of the presentation is to give a brief overview and insight into the families, for personal perspective on the medical diagnosis odyssey.

- First recognized in 1962 by pediatric cardiologist Dr. Jaqueline Noonan, Noonan Syndrome (NS) is a rasopathy and a variably expressed multi-system disorder with an estimated prevalence of 1 in 1,000 to 2,500 births.
- NS is the most common syndrome you have never heard of and is a most often autosomal dominant genetic disorder caused by mutations in several different genes, and there are continuing to be more and more identified.
- Approximately 50% have an affected parent and the risk of passing on the affected gene is 50%; however, there are spontaneous mutations that do occur in individuals.
- Since it is a truly complicated pathway, playing the short You Tube video, *What are the RASopathies (updated)*, provides a much better explanation of the physiology of NS.
- NS has 12 identified mutations, including PTPN 11, which 50% of the patients typically have, and identifying more is a continuous process.
- Current medical guidelines for diagnosis and treatment are available for health care professionals, and the guidelines cover multi-systems and ages and stages of the lifespan.
- The NS medical complications arise throughout the lifespan and affects ear/eye/throat, growth and development, cardiac, lymphatic, hematology, learning/behavior, gastrointestinal, neurological, cancer, orthopedical, and renal/liver, and can be life threatening.
- How NS affects newborn features.
- The details of her child's nine-year medical diagnosis odyssey starting from newborn/infancy, how having a medical background helped, and the "what ifs" of an earlier diagnosis.
- Current research and treatment studies include a RASopathies study on cancer by the National Institutes of Health (NIH) and a MEK inhibitor study on patients with NS.
- Personal efforts include getting studies started, delivering Stay Brave Boxes for NICU, collaborating with Genetics on a family symposium, fundraisers, and speaking at The Noonan Syndrome Foundation Family conferences.
- Not aware of any states that screen for NS during newborn screening and that NS is unlikely to be added due to the number of mutations and the number of mutations continually being discovered.

- Wishing providers could complete more head-to-toe assessments, work with other team members such as Genetics, and to add NS to newborn screening to give families answers and ensure lifelong care.

Member discussion:

- Importance of being reminded about the diagnostic odyssey families and patients travel when conditions are not detected at birth.
- Expressing appreciation for rare disease parents advocating not only for their child's rare condition but the greater rare disease community.
- How common the NICU experience is in the NS community.
- Examples of programs looking to secure funding for diagnosis in the NICU and Pediatric Intensive Care Unit and efforts outside the NBSAC to establish Project Baby Dillo in Texas to secure funding for whole genomic sequencing of NICU babies with unknown etiologies with their parents and to look at their phenotypes to secure a rapid diagnosis.
- How to pick up more babies with NS in the NICU and make a difference on how they work up and provide resources for children.
- The barriers families face in getting insurance to cover testing.
- Due to parent persistency and advocacy for awareness, several conditions have been added to the federal Recommended Uniform Screening Panel (RUSP), which have then been added to the Texas panel.
- The expense of whole genome or whole exome sequencing may be one of the arguments moving forward.
- How families without a medical background have even a harder time navigating the medical system.
- The benefits of early detection and the cost to the public health system.
- If any companies are covering NS testing during clinical trials.

Legislative update

Highlights of the presentation included:

- Multiple bills proposed and some bills passed.
- Passing of House Bill (HB) 2478 changes the language around the Newborn Screening Preservation Account to allow funds to be used, but does not require them to be used, for a seven-day work week.

- HB 2478 also changes language requires an annual report that includes conditions added to the RUSP but not being screened for in Texas and the barriers to implementation.
- An outline has been drafted for the annual report due September 1, which will include four core conditions on the RUSP not screened for in Texas, but that they are in the process of implementing.
- Annual report will also include implementation process status, anticipated go-live date, what funding was used and is planned to be used, and details on and what is required to implement a seven-day work week information.
- Another part of HB 2478 adds a Congenital Cytomegalovirus (CMV) confirmatory test and subsequent hearing screen requirement to Texas Health and Safety Code Chapter 47.

Member discussion:

- Whether the annual report is required to or will address Congenital CMV.
- Rules will have to be added to the Texas Administrative Code regarding the implementation of Congenital CMV testing.
- Since advocates discussed adding conditions such as Niemann-Pick and Duchenne Muscular Dystrophy during the legislative session, if conditions not on the RUSP will be included in the annual report because their intent was to work with the department in the interim on adding conditions not on the RUSP.
- Advocates had the goal of shortening the time and providing the DSHS Laboratory with what is needed to begin screening RUSP conditions as quickly as possible.

ACTION ITEMS

- Ms. Otto will send the You Tube link to program staff to share with the subcommittee.
- Address legislative intent versus interpretation of bill language and whether the annual report needs to include conditions not on the RUSP.

The meeting adjourned at 12:08 p.m.