Newborn Screening Advisory Committee Fall 2023 Meeting

Minnesota Department of Health

October 24th, 2023 | 1:00 p.m. - 3:20 p.m.

Meeting called to order at 1:03 p.m. by chair, Rae Blaylark. Live CART services provided by Megan Stumm, RPR.

Attendance

Advisors Present:

- Rae Blaylark (chair)
- Katie Pfister (vice-chair)
- Jennifer Arveson
- Susan Berry
- Alex Boucher
- Christen Ebens
- Patricia Hall
- Dietrich Matern
- Brooke Moore
- Randal Richardson
- Annamarie Saarinen *(virtually)*
- Kathy Stagni
- Renee Temme
- Marcelo Vargas

Advisors Absent:

- Robert Jacobson *(notified in advance)*
- Courtney Jarboe
- Eva Morava-Kozicz *(notified in advance)*
- Joe Williams

NBS Program Updates

Universal screening for CMV began February 6, 2023.

- In the first 7 months of screening, there have been 91 confirmed cCMV cases, 1 false positive, and 1 false negative case. The screen is good at picking up CMV viral DNA in blood spots.
- A CMV consortium is being coordinated by MDH to provide a space for pediatric infectious disease specialists to come together and talk about specific cases and needs to support families of babies with positive newborn screens. This effort is ongoing.

Recent panel additions: Krabbe disease, MPS II, and GAMT deficiency

- Validation has begun for screening for Krabbe disease in partnership with Mayo Clinical Laboratories and Mayo Clinic's Biochemical Genetics Laboratory.
- External implementation workgroups have been scheduled with neurologists, genetic counselors, bone marrow transplant physicians, parents, and advocates to guide MDH readiness to begin screening for Krabbe disease.
- Goal to begin screening in early 2024, exact date TBD.
- Implementation efforts for MPS II and GAMT will happen later in 2024 when MDH lab has more capacity.

Data dashboards available on Newborn Screening Data Summaries

Committee Business

A nomination was received for Metachromatic Leukodystrophy from Shanna Quimby, co-sponsored by Dr. Paul Orchard and Senator Jeremy Miller.

Vacancies will be opening soon for advisor appointments with four-year terms beginning in January. Requesting recruitment help for: primary care providers, birth hospital representatives, and a nutritionist.

- Applications can be submitted through the Secretary of State website and will be reviewed the first week in December.

Public Comment

Niki Armstrong, CGC – Parent Project Muscular Dystrophy

Marit Sivertson – Mom of a child who has Duchenne Muscular Dystrophy

See separate file for transcript of their public comments.

Presentations

Overview of Duchenne Muscular Dystrophy (DMD) – Dr. Randal Richardson, neuromuscular specialist

Highlights from Condition Readiness Workgroup reviewing evidence related to newborn screening for DMD – McKayla Gourneau, LCGC | MDH NBS Program

MDH Readiness

- Laboratory readiness – Emily Morrison, lab scientist
- Short-term follow-up readiness – Sondra Rosendahl, supervisor
- Longitudinal follow-up – Jennifer Hauser, supervisor

Discussions

History, treatments, outcomes, screening protocol, readiness, cost, MN specific considerations, concerns

- Primary target of screening: Duchenne Muscular Dystrophy – we should strongly consider molecular testing as a part of the screening protocol to minimize false positive results and ensure children identified through screening will be the one's most impacted by DMD.
    - Is there an ethical issue for not reporting out elevated CK-MM cases without a finding on molecular analysis of the dystrophin gene? Screening is not meant to have answers for everything, the focus is on the primary target. Elevations in CK-MM can happen for a variety of reasons – even something as benign as birth trauma. Reporting those elevations without molecular confirmation is also an ethical concern.
    - If molecular analysis is done through the screening program, it would be done through a send out process to an outside lab who will have control over what is being reported out (pathogenic, likely pathogenic, VUS, benign).
        - Concern for clinical utility of variants of uncertain significance (VUSs) – may want to stipulate which results can/should be reported.

- Concern re: access to treatment from an insurance coverage standpoint and date of diagnosis. Newborn screening is not the only time point for boys affected with DMD to be identified and to have maximum benefit.
    - Projected 30% of cases will be eligible for exon skipping therapy that has no age limit for administration (can access at diagnosis in the early newborn period)
    - Corticosteroids are not recommended to be started prior to 2 years of age.
    - Gene therapy – Elevidys – is for boys with DMD between 4-5 years of age. (projected 90% of boys with DMD will be eligible for this treatment)
- Age at diagnosis varies widely. Individuals from lower SES and rural communities are more likely to be diagnosed later after the age of 5. This subset of the population may miss treatment opportunities based on what is currently available without early detection through newborn screening.
- Cost of treatment is expensive. Exon skipping therapy can be upwards of $1 million dollars and the price of Elevidys at this time is $3.4 million. If we add DMD to the newborn screening panel, care should be taken to ensure coverage is available so any child who needs treatment will have access to it.

Decisions Made

Motion by Susan Berry: To recommend adding DMD to the Minnesota newborn screening panel as measurement of CK-MM and if elevated is followed by molecular testing to identify pathogenic or likely pathogenic variants in the dystrophin gene.

Second by Kathy Stagni.

14 in favor, 1 abstain, 3 absent – motion passes.

Closing

Meeting closed at 3:20 p.m.

Next meeting: Tuesday, April 23, 2024 | Location TBD