Meeting Minutes: Advisory Committee for Heritable and Congenital Disorders

October 6, 2020

Minutes prepared by: Holly Winslow
Location: Virtual – using MS Teams

Advisors Present

- Jen Arveson
- Sue Berry
- Rae Blaylark
- Bob Jacobson
- Courtney Jarboe
- Steve Johnson
- Peter Karachunski
- Amy Karger
- Jan Larson
- Cathy Long
- Dieter Matern
- Brooke Moore
- Eva Morava Kozicz
- Steve Nelson
- Katie Pfister
- Teresa Rink
- Annamarie Saarinen
- Kathy Stagni
- Renee Temme
- Marcelo Vargas
- Becca Williams
- Joseph Williams

Advisors Absent

- None

Summary of Decisions Made

- Decision: The October 15, 2019 meeting minutes were approved.
- Decision: Rae Blaylark was approved as Vice Chair and Jan Larson was approved as Chair.

Summary of Action Items

- Action/Assigned to/Due date
    - Further discussion on term limits is needed at a later date.
    - Rae expressed interest in examining refusal data to discover if there are education opportunities to reduce these numbers.

Agenda

- Welcome and Introductions
- Announcements (presented by Mark McCann)
    - General updates– 61 conditions including 2 point of care. Combined about 500 kids benefited.
    - Continuity of operations during COVID presented by Mark McCann. Screening 6 days per week. On call staff 7 days per week. Some staff have moved to working remotely.
- Committee Business
    - October 2019 meeting minutes approved. Motion by Bob Jacobson. Second by Steve Johnson. Passed unanimously.
    - Rae Blaylark has been nominated as Vice Chair. Jan Larson has been nominated as Chair. Motion by Sue Berry. Second by Courtney Jarboe. Passed unanimously.
- Newborn Screening and COVID-19
    - MDH epidemiologist, Tory Whitten, studied whether COVID-19 is impacting blood spot screening in terms of early collections, missing screening, refusal rate, or unsatisfactory specimens. Studied data for over 42,000 babies. Any change due to COVID are small in comparison to babies with normal screening. MDH will continue to monitor these data as well as notification metrics, NICU admission rates, and a correlation between COVID-19 positive mothers and SCID positive NBS results.
        - Slight increase in screens collected early (before 24 hours of age). No change in the distribution of age among specimens collected early.
        - There was an increase in the percentage of unsatisfactory specimens in March-May compared to other 2020 months but not higher than 2019. Unsatisfactory data is highly variable by month.
        - Increase in babies who left the hospital without a blood spot screen collected. Some of the missing screens were due to delays/lost in transit – this data is increased compared to 2019.
        - Refusals are similar between 2019 and 2020. Some evidence of pockets of communities in greater MN with higher refusal rates than average.
    - MDH epidemiologist, Regina Marino, studied the impact of COVID-19 on point of care screening.
        - Early communication was sent out by MDH to emphasize that NBS is essential.
        - Hearing screening
            - Increase in missing hearing screenings and proportion of infants screened dipped in March and April of 2020 while PCP clinics were not seeing healthy patients. Changes in audiology clinic protocols included delayed follow-up and additional documentation needed. Time to diagnosis decreased earlier due to forgoing a repeat screen and going straight to diagnostics.
        - CCHD screening
            - Concerns surrounding shared equipment and early hospital discharges.
            - Increase in babies screened early (before 24 hours of age) in March/April but has since returned to pre-COVID levels.

    - Question from Dieter Matern about using DBS specimens to screen for COVID antibodies. MDH is currently validating this method using our GSP instruments in conjunction with the Infectious Disease Lab at MDH, but not for newborn specimens.
    - Question from Sue Berry about reagent shortages due to shared reagents with COVID testing. MDH has seen some shortages in PPE but the lab keeps at minimum a 3 month supply of all reagents and supplies in house so has not yet been affected by a shortage.
- Sickle Cell Disease Surveillance (presented by Jennifer Hauser and Jay Desai)
    - CDC Sickle Cell Data Collection program has been active since 2015 with a goal to improve quality of life and life expectancy of SCD patients. Objective is to translate population based data to action.
    - MDH received a one year SCDC capacity building grant in order to increase standardization of methods for surveillance and increased understanding of the utility of surveillance data for all stakeholders. No data was collected for the grant but data sharing linkages were created with many existing sources of SCD data including newborn screening. The MDH team was cross-divisional and included a number of external partners and key stakeholders.
    - MDH applied for a three year SCDC implementation grant to continue the progress made during the capacity building phase. Goals are to fully execute data sharing agreements and apply standardized methods to collect SCD data and conduct surveillance and epidemiologic analyses. Aggregate data will be provided to CDC.
    - Kudos from Rae Blaylark to the team on the work completed in year one in spite of COVID-19.
- Potential Condition Consideration (presented by Sondra Rosendahl)
    - No formal nominations are currently being reviewed nationally or in MN at this stage. MDH has a "watch list" of disorders that have potential to be added to the RUSP. Will go through 4 of them that have had clinical/family engagement recently in MN.
    - Krabbe Disease. Clinical information: early and late onset with wide range of progression and severity. Screening method is MSMS and would be multiplexed in MN with our MPS-I and Pompe screening. Second tier testing would be necessary to reduce false positives. Treatment is stem cell transplant but this is not curative. Eight states are currently screening for Krabbe.
    - Metachromatic Leukodystrophy (MLD). Clinical information was reviewed. Screening method is MSMS but cannot be multiplexed with any current testing. Screen is better with urine than blood for this disorder. Second tier testing would be necessary to reduce false positives. Treatment options include gene therapy, enzyme replacement therapy and stem cell transplant for supportive and symptomatic relief. No population based screening currently, but a few states are doing pilot studies.
    - Congenital cytomegalovirus (cCMV). Clinical information: 10/10/80 symptomatic/asymptomatic with hearing loss/asymptomatic. Only window of reliable diagnosis is before 2-3 weeks of life due to high prevalence of CMV in the environment. Screening is PCR – MN is currently doing a pilot study for this screening. Treatment includes increased monitoring for hearing loss for all and antiviral medication for symptomatic children. Targeted screening for babies who do not pass hearing screen is available in about six states as well as select hospitals in MN.
    - Duchenne Muscular Dystrophy (DMD). Clinical information: onset in early childhood and affects males almost exclusively. Screening method is GSP – MN has this instrumentation and there is

an FDA approved reagent kit available. High false positive rates so 2nd tier testing would be needed. Treatment includes corticosteroids but clinical trial for gene therapy is on an FDA fast track. More results are expected in 2021. Pilot studies currently happening in three states.

- Things to note while waiting for official nominations:
  - Will need a fee increase, upgraded/new instrumentation and additional staff, and time to run validations for new testing.
  - Reminder of the new process that was approved to guide the review procedure.
- Question from Sue Berry on the new process regarding how a timeline would be set for new condition consideration. Workgroup would meet after the presentation of expert information and provide guidance to committee on whether the condition is ready for vote or needs more review.
- Question from Annamarie Saarinen on clarifying the exact process for new condition review. MDH clarified that workgroup will not do formal evidence review but have set criteria that must be met for a condition to be added to the panel. The readiness packet would be presented to the full committee from the workgroup to decide on next steps (more time to review/gather data or ready to vote). Approximately a year long process.
- Question from Sue Berry as to weight of readiness criteria for system vs screening program. Former MDH commissioner believed that our Advisory Committee did not need to wait for federal RUSP action but had expertise available to make decisions for the MN panel. MN has a strong advocate community and relationships with legislators to be able to divert legislative actions to the NBS nomination process. MDH reviewed the committee approved critical criteria and supporting factors for nomination process.
- Question from Peter Karachunski on whether MDH waits passively for nominations. The nomination form and packet is available on the MDH website and submitted to the program to be brought to the Advisory Committee.
- Comment from Dieter Matern on Krabbe status. Senator from Winona and Legacy of Angels group have been given the nomination form.

Next Meeting

Date: April 20, 2021
Time: 1-3 pm
Location: Virtual
Agenda items: submit proposed agenda items to Sondra Rosendahl Sondra.Rosendahl@state.mn.us

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