Meeting Minutes: Advisory Committee for Heritable and Congenital Disorders

April 20, 2021

Minutes prepared by: Sondra Rosendahl
Location: Virtual – using MS Teams

Advisors Present

- Jen Arveson
- Rae Blaylark
- Bob Jacobson
- Steve Johnson
- Peter Karachunski
- Jan Larson
- Dieter Matern
- Brooke Moore
- Eva Morava-Kozicz
- Katie Pfister
- Teresa Rink
- Annamarie Saarinen
- Kathy Stagni
- Renee Temme
- Marcelo Vargas
- Joseph Williams

Advisors Absent

- Sue Berry
- Amy Karger
- Becca Williams
- Courtney Jarboe

Summary of Decisions Made

- Decision: the October 6, 2020 meeting minutes were approved.

Summary of Action Items

- Action/Assigned to/Due date
  - Sondra will send out the JAMA publication of the CMV study.
  - Sondra will send out the link to their data webpage once its live.
  - MDH will look into the MN Community Measurement.

Agenda

- Welcome and Roll Call
- Announcements (presented by Mark McCann)
    - Birth rate down by about 5% - COVID impact?
    - Fee increase – active in pursuing newborn screening fee increase; currently charging $135 and looking to add $42. MN also has an off-set cost for support services for the Deaf and Hard of Hearing community of $15 per specimen.
    - Vivian Act
        - Bill for CMV education originally pursued during the Dayton administration
        - CDC, UMN and MDH collaborative research study looking at DBS sensitivity
        - Re-introduced this legislative session; senate version has screening added
        - MDH has provided a fiscal note for this addition
        - Mandate for screening not included in the House version
    - Discussion:
        - Dieter – fee increase but lower birth rate; how does that work? How does it compare to other state programs?
            - Mark – Has been 8 years since the last one, and help to address a foreseeable deficit. MN is in the top 3rd for cost of newborn screening. Some programs are already charging $190ish dollars.
        - Annamarie – Dieter asked her first question. Second question: was the only request for MDH to weigh in on the cost, and has the grant wrapped up?
            - Mark – MDH only provided the fiscal note; CMV is not a cheap test. Study is about a year or two out from completion. Sondra can share the CMV publication with the committee.

- Committee Business
    - October 2020 meeting minutes approved.
        - Motion by Dieter to approve, second by Rae, and passed unanimously
- Rare Disease Advisory Council (presented by Erica Barnes)
    - 2019 governor signed bill for the council to exist
    - Housed at the University of MN, but there are many institutions represented.
    - Reviewed four council pillars
        - Deepen understanding of the needs of the rare community
            - There are unique needs over and above in the rare disease community
            - Just completed a patient survey that have two principal investigators who will be presenting the results of this survey
        - Increase access to and coordination of care
            - Working on developing recommendations and resources for that
            - Transition of care from pediatrics to adults; help figure out strategies
        - Identify strategies for reducing time to diagnosis
            - Some alignment with what your committee is doing

        - Encourage the acceleration of research
            - Many diseases still without an FDA approved treatment
            - Ambitious goal, but we want to focus on the organizational health
    - Raise awareness in our health care system
        - Help institutions understand rare disease as a subset of the population
        - There are silos within the health care system
        - Doing education through the medical schools
        - Statewide survey of primary care providers (barriers they're facing when trying to meet the needs of their patients with rare disease). How do we help bridge the gap between the PCPs and specialists? Explore different use of technologies.
        - Payers
    - Build support for patients and populations
        - Working on a physician contact registry – provide resources and reduce isolation
        - Patient survey – understand barriers
        - Mature patient communities – building out clinical trial readiness tools
        - Support physician-patient relationship – compare the patient and provider surveys to see where the differences are and try to support that relationship
    - Addressing policy impact
        - See ourselves and want to be seen among state agencies as a resource
        - Engage with committees and workgroups
        - Educate legislators and governor's office – were able to encourage that people with rare disease be added to the COVID prioritization list
    - Discussion:
        - Jan – how will the provider registry be disseminated?
            - Erica – will be making it available on our website; one for families and one for clinicians
        - Sondra – who are the "physicians" that will be included in the registry?
            - Erica – will be for primary care providers and specialists. If anyone would like to be added to this registry, she can send the google form
        - Peter – would like to connect with Erica about their transition of care activities at the UMN neuromuscular clinic
    - Sondra has my contact information if you'd like to have any follow-up conversations
        - Results of their survey will be available mid-May and publicly available for anyone interested in seeing those results
- Family Story – Myra's Journey (given by mother of Myra, Kelsey Herickhoff)
    - Kelsey shared how and when they heard about their daughter's newborn screening result, learning that their daughter had SMA type 1, and their treatment journey. Myra is now 2 years old and loves to dance, be outside, and jump on their trampoline.
    - Q&A:
        - Jen – did I hear that right that she was 10 days old when you got the screening result?
        - Kelsey – Yes, I have been working with the birth hospital pre-COVID to improve this. I have met with other families who had their results at 4 days old.

        - Mark – screen was received on Friday, testing began on Monday with notification on Tuesday
        - Dieter – Also concerned; MDH does scorecards for submitters, perhaps we can look at those; press could be very bad for them.
        - Kelsey – would like more help in getting the hospital to do something about it
        - Bob – has anyone involved ICHSI and the MN Community Measurement?
        - Mark –great suggestion by Bob; perhaps our QA officer and I can look into this further.
- Look Back: Spinal Muscular Atrophy (presented by Dr. Peter Karachunski and Tory Whitten)
    - Background on cause of SMA and the various types (type 1 being the most severe)
    - Reviewed treatment options available to patients
        - Zolgensma approved in May 2019; error on slide that says May 2018
    - MDH screening data was presented
    - Process for follow-up and outcomes from his center were also shared
    - Discussion:
        - Dieter – treatment initiation needs to happen faster. Are you working with other state's to develop guidelines that might be needed to work with insurance providers to improve their approval timing?
            - Peter – good idea to collectively discuss this to get them to change their policies
        - Dieter – MDH used to provide annual summaries – would like to hear more about what data we have on all of our conditions
            - Sondra – Tory and I are working on getting this type of data on our webpage. Data is ready to go and just need to work with our web person to get it added. I will send out the link once it's live.
        - Annamarie – did you have a more specific talk or recommendation for your bullet point "earlier NB screening sample collection" or is it more about reducing the time lag
            - Peter – as I said some of this may not be doable, but understands it would complicate things logistically
        - Annamarie – Is there data at present in MN or otherwise to say from how long to point of notification to approval and what the variables are?
            - Peter – we probably could get this data. Time to treatment does not reflect approval. Able to work with Biogen to request bridge therapy before we get approval.
            - Annamarie – but is that unique to you?
            - Peter – I don't think so, believes it's part of their policy. All who treat know about it. I won't wait for gene therapy approval before starting bridge therapy.
            - Annamarie – great to do more work with the follow-up workgroup at the federal level as she really enjoyed listening to this session
        - Jen – Is ten days reasonable? Do we need to be doing more education?
            - Mark - in regards to turnaround time, if we see batching, they get a phone call. Twice per year, submitters also get the QA reports that show their performance.
        - Mark – our lab operates screening SMA using a 5 day per week model because it wasn't considered time-critical at the time of approval. Should we be going to 6 days?

        - Peter – seems reasonable to discuss this further; would help with their planning and not necessarily that they would need to go to the ER
    - Dieter – Annamarie, you should ask for ACHDNC to reconsider time-criticalness. With no false positive results, there should be no reason to not get them to treatment ASAP.
        - Annamarie – Agree. Dieter and Peter, let's reconnect on this prior to the May ACHDNC meeting.
    - Mark – of note, we pay for UPS overnight shipping
        - Dieter – so there is no excuse for the submitters to not get the specimens to MDH in a timely manner
    - Amy – we also match our specimens with birth certificates, and we are proactively reaching out to make sure that those specimens are getting to us as soon as possible.

Next Meeting

Date: October 26, 2021
Time: 1-4 pm
Location: TBD
Agenda items: submit proposed agenda items to Sondra Rosendahl Sondra.Rosendahl@state.mn.us

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