November 8, 2018

Darren Michael, PhD
Newborn Screening Program Manager
8100 E. Lowry Boulevard
Denver, CO 80230

RE: Proposed Newborn Screening Rules, 5 CCR 1005-4

Dear Dr. Michael,

On behalf of our coalition of child health advocates and healthcare providers, thank you for your continued willingness to partner with the stakeholder community to strengthen the newborn screening program. We are writing to comment on the proposed rules that will be considered during the December rulemaking hearing. We would like to recommend the following considerations which we have divided into two areas, lab functions and newborn screening programmatic functions:

Lab functions

- *Cutoffs for medical conditions should be referenced in rules*
    - We urge the Department to add a reference in rule regarding their role in working with stakeholders and medical professionals to ensure that there is a periodic review of cutoff values. This could be added in the quality section of the regulation.
    - Recommended language: The Department will ensure continuous quality improvement by scheduling periodic review of lab cutoff values, with input from stakeholders and medical professionals.

- *Second-tier testing at the lab*
    - In the recently revised CRS 25-4-1004.5(2)(b), the statute was amended to ensure the newborn screening fees are sufficient to cover the cost of implementing second tier testing at the Department in order to address the number of false-positive test results. We appreciate that CDPHE has been working on this issue for a number of years. As enumerated in the new law, this issue is critical to facilitate greater cost-efficiency in the system and to reduce the number of families who are impacted by false-positive test results. We urge the Department to ensure a mechanism for updating stakeholders and evaluating second-tier testing considerations, in compliance with the statutory obligations.
    - Recommended language: Please see enclosed recommended changes in the quality section of the rules. Additionally, we recommend language that, "the Department will continue to analyze second-tier testing capacity at the laboratory to address false-positive test results. The Department will periodically provide updates to stakeholders and the medical community to evaluate progress on second-tier testing."

- *"Time-critical condition" and "time-sensitive condition" definitions*
    - We appreciate that "time-critical" and "time-sensitive" definitions have been added to the rules, however these terms are not used again in the rules. Instead, the later content of the rules uses "urgent" and "non-urgent" results. To ensure uniformity and clarity, we recommend using "time-critical" and "time-sensitive" in place of "urgent"

and “non-urgent” in alignment with nationally recognized terms. The conditions associated with these terms are not identified in rule. While borderline results may create some ambiguity for these terms, the Department may want to consider defining which conditions fit these profiles so that the timelines for follow-up are clear to primary care and specialty providers and families.

- *Designee*
    - The rules use the term “designee” regarding follow-up with providers and families. It is unclear who is intended to be considered a designee and it would be helpful to define this term in the definitions section to ensure clarity about who has the rights and responsibilities to take on these duties.

- *Supplementary criteria for adding new conditions*
    - In the recent stakeholder meeting, a separate draft document was proposed for adding language regarding the consideration of new conditions in the newborn screening program. We believe these criteria go beyond the statutory authorization of the Department and the Board of Health. The statute clearly delineates four components for consideration of a new condition and we are not aware of an allowance for additional criteria. The current language appears to commit the program to identifying data that are unlikely to be easy to obtain and which the lab is not in a position to obtain. For example, the incidence in Colorado will not be known for most conditions and can’t be easily obtained. The same is true for the sensitivity and specificity language. The added criteria could make it nearly impossible to add new conditions on the newborn screening panel in Colorado.
    - <u>Recommended language:</u> We recommend against the addition of any new criteria beyond the statutory authorizations currently included in the law.

Newborn screening programmatic functions

- *Elimination of quality improvement and education components in the rules*
    - As shared in the stakeholder meeting, we are very concerned about the elimination of language related to quality improvement and education in the statute. This is a critical foundation for a successful newborn screening program and must have regulatory support for effectiveness. In CRS 25-4-1003, it is clear that the legislature intends for the newborn screening program to be “carried out under adequate standards of supervision and quality control” and further, that education programs must be delivered to increase the public’s understanding of newborn screening and to establish systems for recording information to include in genetic counseling and education programs. As such, we strongly recommend ensuring the quality and education language is added back into the rule.
    - <u>Recommended language:</u> Please see enclosed recommendations.

- *Program evaluation is critical and currently missing from rules*
    - Beyond quality improvement efforts within the laboratory and within other healthcare provider domains, it is critical to have a program evaluation component included in the rules. This should include sharing regular updates with stakeholders including the medical community and advocates. Please see enclosed example of Ohio’s transparency with program evaluation information. Typically, states provide, at a minimum, annual

performance reviews to provide transparency on metrics such as timeliness of blood spot submissions, information specific to individual birthing facilities, false-positives, false-negatives, aggregated confirmatory diagnoses, and other measures. The goal of newborn screening data reporting and evaluation should be to set clear expectations and coordinate outcomes. This is a current gap in Colorado's newborn screening program.
    - Recommended language: Please see enclosed recommended language.

- *6-month case closure*
    - We recommend updating the language regarding the proposal that follow-up services shall not be provided after 180 days. Testing and working with families may still be ongoing at 6 months. If a timeline is necessary to define in rule, we urge a one-year timeframe to allow time to track those cases and determine more conclusive results that can be reported.
    - Recommended language: We recommend moving this language to another area of the rule and encourage language that articulates, "For purposes of program evaluation the Laboratory shall work with providers to collect information regarding final diagnosis of infants with positive newborn screening results and will maintain a mechanism for providers to report missed cases. Cases may be closed as 'lost to follow up' if the family cannot be located or chooses not to be contacted for a period of one year. At the discretion of the Laboratory and provider, cases may be kept 'open' longer than one year if there is programmatic value to so doing."

- *Advisory council*
    - We recognize there is not a formal advisory council described in the law or in current rules. However, we urge consideration of some language in the rule to describe the important partnership and functions among the Department, the medical community, parents, and child health advocates. These advisory relationships are critical for all to share responsibility in pursuing improvements to the newborn screening system in Colorado and we think it is valuable to include a reference that provides some recognition for this collaboration and more formal feedback mechanisms among all entities responsible for the care of newborns.

Thank you again for your willingness to consider our input on these rules. We look forward to continued dialogue and partnership to strengthen the newborn screening program in Colorado.

Sincerely,

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