COLORADO
Department of Public Health & Environment

To: Members of the State Board of Health

From: Dr. Darren Michael, Newborn Screening Business and Strategy Manager, Laboratory Services Division

Through: Randy Kuykendall, RK
Interim Administrative Director, Laboratory Services Division

Date: December 19, 2018

Subject: **Rulemaking Hearing**
Proposed Amendments to 5 CCR 1005-4, *Newborn Screening and Second Newborn Screening* with a rulemaking hearing on December 19, 2018

When an infant is born in Colorado, two blood specimens are collected and forwarded to the Department for the testing of 37 rare genetic and metabolic conditions. The initial newborn screening specimen is taken by the hospital, birthing facility, or midwife within 48 hours of birth; the second specimen is collected by the infant's pediatrician approximately 8 to 14 days after birth for certain conditions identified in the rules. In the event of a positive result, a specialist on the condition reaches out to the family of the infant to arrange for follow-up services and/or additional testing.

At present, the *Newborn Screening and Second Newborn Screening* rules perform the following functions:

a) Define key terms,
b) Establish procedures for the collection and submission of blood spot specimens for testing,
c) Establish procedures for testing of specimens and reporting of results,
d) Establish requirements for quality control and education, and
e) List conditions covered by the initial and second newborn screening panels.

Together, these definitions, procedures and requirements establish roles and responsibilities for the metabolic testing portion of Colorado's Newborn Screening Program. Historically, the majority of the rule was promulgated through the State Board of Health, with the Executive Director having rulemaking authority over aspects of second newborn screening.

In 2018, House Bill 18-1006, *Infant Newborn Screening*, updated and expanded the statutes related to newborn screening in Colorado. Though the statutes were reorganized, the rulemaking authority that serves as the basis for the current rules is largely unchanged. HB 18-1006 did transfer the Executive Director's rulemaking authority to establish the standards for second specimen testing to the Board of Health. To effectuate the change the Department requests the Executive Director repeal the existing Executive Director rules and that the Board of Health adopt the second newborn screening rules as modified in this rule making.

The one substantive change that relates to rulemaking is HB 18-1006 expanded the Board of Health's rulemaking authority and require rules related to follow-up services. The proposed changes implement the new statutory directive. Last, the proposed revisions implement the results of the Department's regulatory efficiency review that occurred pursuant to E.O. D 12-002 and Section 24-4-103.3, C.R.S. This includes updates to align with current statutory language and statutory citations.

The proposed revisions do not address the newborn hearing screening rulemaking requirements in HB 18-1006. These rules are being developed by the Department's Center for Health and Environmental Data. It is anticipated that these will come to the Board of Health in early 2019. Similarly, this rulemaking does not add new conditions or modify the list of conditions on the panel.

The Department has received stakeholder feedback since the time of the request for rulemaking presentation. Feedback has been reviewed and incorporated into the proposed rule as appropriate. The vast majority of the revisions since the request for rulemaking ensure consistent use of the terminology and elaborate upon the explanation previously provided. A substantive change was made to Section 4.2.3 (Board of Health rulemaking). Along with revising the timeframe for follow-up to better align with the definitions, language was added to recognize that time is of the essence when making referrals and the end date for referral services was extended from 180 days to 365 days as this allows follow-up services for children that meet the definition of "exceptional circumstances."

Notable changes to the rulemaking packet since the request for rulemaking presentation are highlighted in yellow.

STATEMENT OF BASIS AND PURPOSE
AND SPECIFIC STATUTORY AUTHORITY
for Repeal of Executive Director Second Newborn Screening Rules
and Amendments to
5 CCR 1005-4, Newborn Screening and Second Newborn Screening
promulgated by the Board of Health

Basis and Purpose.

The *Newborn Screening and Second Newborn Screening* rules perform the following functions:

a) Define key terms,
b) Establish procedures for the collection and submission of blood spot specimens for testing,
c) Establish procedures for testing of specimens and reporting of results,
d) Establish requirements for quality control and education, and
e) List conditions covered by the initial and second newborn screening panels.

Together, these definitions, procedures and requirements establish roles and responsibilities, for the genetic and metabolic testing portion of Colorado's Newborn Screening Program.

The following changes to the rules are being proposed:

1) The Department proposes several modifications and additions to definitions in Section 1.2 of the rules. The new definitions of "Named Submitter" and "Birthing Facility" clarify roles and responsibilities, and align with new statutory language. The new definition of "Exceptional Circumstances" acknowledges situations, such as refugee populations, where timely collection may be challenging. It is current practice for the Department to accept specimens from children up to 365 days of life when the child can still benefit from screening. "Newborn" has been defined using guidance from the World Health Organization. Several definitions such as "Screen Negative," "Screen Positive," "Time-critical Screen Positive," "Time-sensitive Screen Positive Result," "Time-Critical Condition," and "Time-Sensitive Condition" are taken from or adapted from the Clinical and Laboratory Standards Institute and the Society for Inherited Metabolic Disorders. "Follow up" is also defined as described below in item 4b. Many of these definitions are introduced as part of implementing new rules required under HB 18-1006.

2) Quality control and education standards are communicated in the current Newborn Screening Regulations and Implementation of Second Newborn Screening sections. Upon review of the statutory authorization for rulemaking, this language has been removed from the initial newborn screening rules as the statute does not require or authorize board of health rules directing the Department to provide quality assurance plans and education.

   Under the Executive Director's administrative responsibilities, rulemaking to establish standards for quality control and information about the programs is authorized pursuant to Section 25-4-1003(2)(a) and (f), C.R.S. The Executive Director is authorized to promulgate rules; however, the Department determined that rules are

not required as this activity can be managed administratively. Though removed from the rule when repealing the Executive Director rules, the Department's quality assurance and educational activities will continue.

The department agrees with stakeholders that quality control and education is an important programmatic activity for the Laboratory Services Division Newborn Screening Program. The Department studied whether Executive Director rulemaking on this topic was merited and found the rules to be unnecessary, and possibly redundant or restrictive. Regulatory review efforts are directed by Executive Order D 12-002 and Section 24-4-103.3, C.R.S.; these legal mandates direct the Department to not create rules unless they are necessary. Here the rules are not necessary. The activities covered under these proposed rules represent operational aspects of the Colorado Newborn Screening Program, which are directed through policies and procedures established by CDPHE's Executive Director. Rules are not typically used to direct how the Department conducts its business as this is directed by the Executive Director as part of the Executive Director's management and oversight of Department programs. The Department is committed to quality control and education and has venues to communicate its efforts with interested stakeholders.

The Department recognizes that Section 25-4-1004.5(3)(b), C.R.S., authorizes the Board of Health to promulgate rules governing supervision and quality control standards for second specimen testing. To harmonize Board of Health rulemaking authority and Executive Director rulemaking authority, the Department has applied the commonly accepted practice that Board of Health rules communicate public policy and direct those with whom the Department works while Executive Director rules typically direct Department programs or Department program activities. (CDPHE also has Chief Medical Officer rulemaking which is often comparable to Board of Health rulemaking.) The Department recognizes that the Board of Health can promulgate rules governing supervision and quality control standards for named submitters who provide second screen specimens. The Department does not recommend the Board of Health promulgate additional quality control and supervision rules. The second newborn screening rules embed quality control. Examples include hygienic practices, sufficient drying time and timely submission. At this time, the Department does not see additional quality control or supervision standards as being necessary. The rules are the minimum necessary for named submitters to accomplish the program requirements. The Department will monitor this; if a need arises to establish standards in rule, the Department will assess whether the standards need to apply to initial and second screening, which lends itself to Executive Director rulemaking, or Board of Health rulemaking for second screening.

3) In Sections 2.2.1.2 and 3.2.2, drying times appears as a range from three (3) to four (4) hours. The reason for providing a range is to ensure named submitters are aware of the minimum acceptable drying time, as well as to maintain awareness that specimens should not be left indefinitely to dry, and instead, submitted within 24 hours following

collection. The proposed rule also indicates that the specimen will dry "horizontally." This is a clarifying language sought by stakeholders that is current practice.

4) The substantive changes to Section 3 include new language to support the collection of a second newborn screening specimen at newborn well child visit. The term 'newborn well child' visit is used widely by the pediatric medical community, who applies this rule. Also, the term 'post partum' was removed to avoid confusion for obstetrics and gynecology medical providers, who typically see mothers approximately six weeks after delivery for what is considered a post partum exam in those medical communities.

5) Section 4 of the proposed rule is written to reflect recent changes to statute through House Bill 18-1006.

   a. The rules now clarify that the state newborn screening laboratory operates six days per week, as required by statute.

   b. Definitions and requirements related to follow-up services are now included.

      Section 25-4-1004.5(2)(c), C.R.S. states,

      The state board [of health] shall promulgate rules to establish and maintain appropriate follow-up services on positive screen cases in order that measures may be taken to prevent death or intellectual or other permanent disabilities. The follow-up services must include

      [i] identification of newborns at risk for genetic conditions,
      [ii] coordination among medical providers and families,
      [iii] connecting newborns who screen positive to timely intervention and appropriate referrals to specialists for follow-up and diagnostic testing, and
      [iv] additional duties as determined by the [Colorado Department of Public Health and Environment].

      (Numbering, in the form of [i]-[iv], has been added to statutory text to aid with analysis.)

      To implement HB 18-1006, the proposed rule establishes follow-up services for any positive screen result with the proposed language in Section 1.2 and Section 4.2 through 4.2.5.

      - The rule defines follow-up services in relation to time-critical and time-sensitive positive screen results; this language implements [i]. The proposed rule also establishes the time frame in which follow-up services may occur.

- The proposed rule requires coordination among medical providers and families as a newborn moves from birthing procedures to pediatric care; this language implements [ii].

- The proposed rule includes referral and confirmatory testing; this language implements [iii]. In addition, to manage false-positives, the rule recognizes that the Department may require repeat or confirmatory testing prior to initiating referral services. Rule 4.2.3 was clarified to recognize that time is of the essence when making referrals while also acknowledging that if delays occur due to confirmatory testing or difficulties contact the parent or legal guardian, the department may extend the period in which referral occurs. The rule sets the range; however, specific timing requirements within that range will be communicated in any contracts. The range aligns with the definition of newborn and acknowledges exceptional circumstances which allow for screening up to 364 days from birth. For some children falling into the exceptional circumstances category, follow-up services may be appropriate.

- The proposed language then communicates the Department will monitor participation in follow-up services; this language implements the discretionary language at [iv].

The Department acknowledges that Newborn Screening is one of many services Colorado provides. Services offered through the Colorado Department of Human Services or the Department of Health Care Policy and Financing, such as early intervention services and services for individuals with intellectual or developmental disabilities may also be available to newborns, and families, who test positive for a condition. The statute references medical providers and specialists, this rule making language focuses on health care services. Through implementation and on-going monitoring, the Department will consider if follow-up services can and should include services offered through our sister agencies.

Also note that the on-going monitoring of the newborn is not included in the proposed definition of follow-up services. For the purpose of the proposed rule, monitoring is limited to evaluation of the newborn screening program. Other programs such as the Colorado Department of Public Health and Environment's Colorado Responds to Children with Special Needs (CRCSN), the state birth defects registry, already monitor information on children diagnosed with conditions that are part of the newborn screen. CRCSN conducts population-level health surveillance, including relevant medical record review and examination of risk factors for individuals up to age 3, on cases with these conditions and other birth defects. In addition to these health surveillance

activities, CRCSN also provides parents with information on community resources for their children. To extend the newborn screening monitoring of individuals beyond what is needed to perform effective screening and follow-up services is an unnecessary duplication of services.

6) Statutory citations, terminology, clarifying edits and formatting changes to improve readability are proposed throughout the rule. The terms mother and father are replaced with the gender neutral terms "parent(s)" and "legal guardian(s)." Stakeholders requested that the term "contractor" rather than "designee" be used in Section 4 as it is a clearer and more relatable term. Upon review of the statute, the Department determined that "contractor" can be used and thus, this change appears in Section 4.

This rulemaking does not propose new conditions or otherwise modify the current list of conditions included on the newborn screening or second newborn screening panel. The specific criteria to be used by the Board of Health for evaluating new disorders for inclusion in newborn screening are stated in statute, C.R.S. 25-4-1004(1)(c) is unchanged. Pursuant to Section 25-4-1004(1), C.R.S., the Board of Health criteria for adding additional conditions remain:

(I) The condition for which the test is designed presents a significant danger to the health of the infant or his family and is amenable to treatment;

(II) The incidence of the condition is sufficiently high to warrant screening;

(III) The test meets commonly accepted clinical standards of reliability, as demonstrated through research or use in another state or jurisdiction; and

(IV) The cost-benefit consequences of screening are acceptable within the context of the total newborn screening program.

HB 18-1006 authorized the Department to take preliminary steps such as a space study and cost assessment to determine the viability of adding new conditions. These actions are occurring. In addition, as previously directed by the Board of Health, the Department is developing a methodology that will be used to determine whether a condition should be recommended to the Board of the Health for addition to the screen schedule. The proposed revisions in this rule establish the structure that will inform that analysis. The proposed rules are predicated on a determination that screening is appropriate based upon state statute or Board of Health rules.

Specific Statutory Authority.

These rules are promulgated pursuant to the following statutes: Sections 25-4-1004 and 25-4-1004.5, C.R.S.

SUPPLEMENTAL QUESTIONS

Is this rulemaking due to a change in state statute?

___X___ Yes in part, the bill number is _HB18-1006_. Rules are ___ authorized
_X_ required.

_____ No

Does this rulemaking incorporate materials by reference?

_____ Yes ___ URL or ___ Sent to State Publications Library

__X____ No

Does this rulemaking create or modify fines or fees?

_____ Yes

___X___ No

The fees for newborn screening are established administratively by the Department. The fees are discussed in this rulemaking packet for informational purposes only.

Does the proposed rule create (or increase) a state mandate on local government?

_X__ No. This rule does not require a local government to perform or increase a specific activity for which the local government will not be reimbursed.

___ No. This rulemaking reduces or eliminates a state mandate on local government.

___ Yes. This rule includes a new state mandate or increases the level of service required to comply with an existing state mandate, and local government will not be reimbursed for the costs associated with the new mandate or increase in service.

The state mandate is categorized as:

___ Necessitated by federal law, state law, or a court order
___ Caused by the State's participation in an optional federal program
___ Imposed by the sole discretion of a Department
___ Other: _____

Has an elected official or other representatives of local governments disagreed with this categorization of the mandate? ___Yes ___No
If yes, please explain why there is disagreement in the categorization.

Please elaborate as to why a rule that contains a state mandate on local government is necessary. NA

REGULATORY ANALYSIS
for Repeal of Executive Director Second Newborn Screening Rules
and Amendments to
5 CCR 1005-4, Newborn Screening and Second Newborn Screening
promulgated by the Board of Health

1. A description of the classes of persons affected by the proposed rule, including the classes that will bear the costs and the classes that will benefit from the proposed rule.

These rules have the greatest impact on all Colorado newborns, of which there is roughly 67,000 a year that are screened, their families, and adult patients with rare congenital disorders. The screen allows for infants to be identified for early intervention and support medical services. These rules impact birthing facilities, midwives, reference laboratories (e.g. Quest, LabCorp), pediatrician's offices, and family medicine offices as the providers who participate in the specimen collection, clinical specialists that currently contracted with Department to provide follow-up services, of which there are about twenty. The rule is of interest to patient advocacy groups (e.g. March of Dimes, Cure SMA, etc.) who advocate for individuals who test positive for one of the various conditions.

A. Identify each group of individuals/entities that rely on the rule to maintain their own businesses, agencies or operation, and the size of the group:

The entities and individuals that are involved in the taking of the specimen who rely on the rules for clarity and certainty include:

- birthing facilities, which include hospitals and birthing centers (over 100 such are entities are licensed by the Department),
- midwives who participate in births that take place outside of a birthing facility (roughly 70 such births take place a year, midwives are certified by the Department of Regulatory Affairs),
- Quest and LabCorp as reference laboratories to the Department, and
- physician offices that provided care to newborns.

B. Identify each group of individuals/entities interested in the outcomes the rule and those identified in #1.A achieve, and if applicable, the size of the group:

There are about twenty clinical specialists that currently contracted with Department to provide follow-up services to newborns who test positive for a condition. Changes to the rules impact these groups in how they provide their services to the families and newborns. Adults with the conditions and advocacy groups are members of our stakeholder process; they are interested in the newborn screening process to help ensure that newborns are connected to specialized care in a timely manner.

C. Identify each group of individuals/Entities that benefit from, may be harmed by or at-risk because of the rule, and if applicable, the size of the group:

The screening process identifies between 80-100 newborns per year for a critical condition out of the roughly 67,000 newborns screened.

2. To the extent practicable, a description of the probable quantitative and qualitative impact of the proposed rule, economic or otherwise, upon affected classes of persons.

A. For those that rely on the rule to maintain their own businesses, agencies or operations:

Describe the anticipated favorable and non-favorable non-economic outcomes (short-term and long-term), and if known, the likelihood of the outcomes:

The proposed rules modernize the metabolic portion of the newborn screening program. The majority of changes are clarifying and updates to align with the current statute and the current operation of the Colorado Newborn Screening Program.

Prior to HB 18-1006, the Newborn Genetics Cash Fund balance was $6.3 million. After the General Assembly increased spending authority, the Executive Director authorized the Laboratory Services Division to increase the current fee from $92 to $111.00. The fee adjustment will accrue an additional $1.3 million to support the newborn screening program generally and fund the activities delineated in HB 18-1006, as well as the anticipated implementation of new conditions. (The legislation also authorized additional funding for newborn hearing screening; however, that is not included in this rule.)

Favorable non-economic outcomes:

Timely screening and follow-up services that bridge the birth to pediatric services benefit the medical providers involved. Medical providers benefit directly from the follow-up services covered by the newborn screening program involved, as these services provide immediate access to experts in the relevant condition for which the child screened positive and work with the family and follow up services to improve outcomes for the identified babies.

Unfavorable non-economic outcomes: None expected

Anticipated financial impact:

Anticipated Costs:	Anticipated Benefits:
Description of costs that must be incurred. • None expected. Description of costs that may be incurred. • None expected.	Description of financial benefit. • To the extent clarifying the language improves provider practice and error rates, a cost and times saving may occur.
Cost or cost range. $_____None_____ or ___ No data available.	Savings or range of savings. $_____ or _X__ No data available.
Dollar amounts that have not been	Dollar amounts that have not been

captured and why: NA	captured and why: NA

Local Government Impact: NA

B. For those that are affected by or interested in the outcomes the rule and those identified in #1.A achieve.

Describe the favorable or unfavorable outcomes (short-term and long-term), and if known, the likelihood of the outcomes:

The proposed rules modernize the metabolic portion of the newborn screening program. The majority of changes are clarifying and updates to align with the current statute and the current operation of the Colorado Newborn Screening Program.

Favorable non-economic outcomes:

Clarifying the existing processes and establishing the standards for follow-up services is of interest to entities that may serve as the Department's contractor and organizations that advocate for patients and families.

Unfavorable non-economic outcomes: None expected.

Any anticipated financial costs monitored by these individuals/entities?

The Department does not anticipate a cost increase to the providers who are involved in obtaining specimens due to the proposed language changes.

Any anticipated financial benefits monitored by these individuals/entities?

Greater operational efficiency of the metabolic portion of the newborn screening program might lower costs for these individuals due to fewer requests tied to unsatisfactory specimens.

C. For those that benefit from, are harmed by or are at risk because of the rule, the services provided by individuals identified in #1.A, and if applicable, the stakeholders or partners identified in #1.B.

Describe the favorable or unfavorable outcomes (short-term and long-term), and if known, the likelihood of the outcomes:

By defining follow-up in rule, the Department is providing clarity about the boundary between newborn screening services and traditional medical services. These rules help to explain the reporting of results according to whether the results are screen negative, time-sensitive screen positive, or time-critical screen positive results and ensure timely processing for newborns and their families.

Early testing allows for early intervention and support to allow the newborns to maximize their chances for success in life. This also gives their families the support they need as they are addressing an emotional fraught issue. The proposed rule

changes clarify the newborn screening process, and establish the process to connect newborns and their families with specialized care and support.

Financial costs to these individuals/entities:

No new costs are expected for these entities and individuals. Any parent or legal guardian may choose to not participate.

Financial benefits to or cost avoidance for these individuals/entities:

For newborns with a screen positive result, the benefit of the proposed rule is that parents, legal guardians and a newborn's medical provider(s) have important information in a timely manner. The costs for early interventions and treatment vary and can be significant; however, these services prevent death, permanent disability, or potentially more significant medical costs if newborn screening results are not provided or are delayed.

3. The probable costs to the agency and to any other agency of the implementation and enforcement of the proposed rule and any anticipated effect on state revenues.

A. Anticipated CDPHE personal services, operating costs or other expenditures:

The newborn screening program statutory mandates are better resourced with the increased appropriation. The rule elaborates and establishes standards related to those mandates; the rule does not drive additional costs.

The newborn screening fee was increased from $92/child to $111/child on July 1, 2018. This was the first fee increase in seven fiscal years. The fee is not established by the Board of Health, but is set administratively through the Executive Director. This rulemaking does not include fees or fee increases. The Department will continue to monitor the fee as it evaluates the programmatic infrastructure and costs associated with improving or expanding the services offered.

B. Anticipated personal services, operating costs or other expenditures by another state agency:

The Department of Health Care Policy and Financing will have costs associated with increased capitation payments (FY19 $117,900; FY20 $123,200; FY21 $139,300) for Medicaid and the Children's Basic Health Plan (CHP+).

It was not anticipated that HB 18-1006 would increase the number of Colorado newborns identified as persons with an intellectual or developmental disability and thus, no additional costs were identified in this area.

Anticipated Revenues for another state agency:

These costs are funded through a variety of funding streams and are to be addressed through the annual budget process.

4. A comparison of the probable costs and benefits of the proposed rule to the probable costs and benefits of inaction.

Check mark all that apply:
_X_ Inaction is not an option because the statute requires rules be promulgated.

_X__ The proposed revisions are necessary to comply with federal or state statutory mandates, federal or state regulations, and department funding obligations.

_X_ The proposed revisions appropriately maintain alignment with other states or national standards.

_X__ The proposed revisions implement a Regulatory Efficiency Review (rule review) result, or improve public and environmental health practice.

_X_ The proposed revisions implement stakeholder feedback.

_X_ The proposed revisions advance the following CDPHE Strategic Plan priorities:

Goal 1, Implement public health and environmental priorities Goal 2, Increase Efficiency, Effectiveness and Elegance Goal 3, Improve Employee Engagement Goal 4, Promote health equity and environmental justice Goal 5, Prepare and respond to emerging issues, and Comply with statutory mandates and funding obligations

Strategies to support these goals:
___ Substance Abuse (Goal 1)
___ Mental Health (Goal 1, 2, 3 and 4)
___ Obesity (Goal 1)
___ Immunization (Goal 1)
___ Air Quality (Goal 1)
___ Water Quality (Goal 1)
_X__ Data collection and dissemination (Goal 1, 2, 3, 4 and 5)
___ Implements quality improvement or a quality improvement project (Goal 1, 2, 3 and 5)
___ Employee Engagement (career growth, recognition, worksite wellness) (Goal 1, 2 and 3)
_ _ Incorporate health equity and environmental justice into decision-making (Goal 1, 3 and 4)
___ Establish infrastructure to detect, prepare and respond to emerging issues (Goal 1, 2, 3, 4, and 5)

___ Other favorable and unfavorable consequences of inaction:

5. A determination of whether there are less costly methods or less intrusive methods for achieving the purpose of the proposed rule.

Rulemaking is proposed when it is the least costly method or the only statutorily allowable method for achieving the purpose of the statute. The specific revisions proposed in this rulemaking were developed in conjunction with stakeholders. The benefits, risks and costs of these proposed revisions were compared to the costs and benefits of other options. The proposed revisions provide the most benefit for the least amount of cost, are the minimum necessary or are the most feasible manner to

achieve compliance with statute. The proposed rules for follow-up services afford the Department some discretion in designing and managing those services.

6. Alternative Rules or Alternatives to Rulemaking Considered and Why Rejected.

See the responses at #4 and #5 above.

7. To the extent practicable, a quantification of the data used in the analysis; the analysis must take into account both short-term and long-term consequences.

The program's laboratory information management system was used to estimate the number of newborns screened by the program.

The Department solicited feedback at all six of the current follow-up clinics (hemoglobinopathies, congenital hypothyroidism, congenital adrenal hyperplasia, cystic fibrosis, severe combined immunodeficiency, and inherited metabolic disorders). The Department has monitored the feedback as these entities have subject matter expertise and a commitment to newborn screening; however, as entities that receive funding to perform these services, there is a conflict or perceived conflict of interest. Their expertise informed the proposed rules.

The Department also reached out to a neonatologist in Colorado Springs, and a pediatrician in Grand Junction.

The Department has also used the survey results to open a dialogue with birthing facilities and individuals involved in births that occur outside of a birth facility. Outreach to this community continued. The survey results also informed the proposed rule language.

The Department received written feedback from Children's Hospital of Colorado and the Colorado Midwives Association regarding our originally proposed rules. The current proposed rules reflect significant changes made to incorporate this input.

The Department has also reviewed the following documents:

1. Clinical and Laboratory Standards Institute (CLSI). Newborn Screening Follow-up; Approved Guideline—Second Edition. CLSI document NBS02-A2 (ISBN 1-56238-875-4 [Print]). Clinical and Laboratory Standards Institute, 950 West Valley Road, Suite 2500, Wayne Pennsylvania 19087.
2. APHL Presentation by Dr. Susan Tanksley
3. "Timeliness of Newborn Screening: Recommendations " https://www.aphl.org/conferences/proceedings/Documents/2015/Annual-Meeting/26Tanksley.pdf
4. Society for Inherited Metabolic Disorders "SIMD Position Statement: Identifying abnormal newborn screens requiring immediate notification of the health care provider." https://www.simd.org/Issues/SIMD%20NBS%20Critical%20Conditions%20policy%20statement.pdf
5. Dr. Joe Orsini, "Overview of Cutoff Determinations and Risk Assessment Methods used in Dried Blood Spot Newborn Screening." ACHDNC, February 8, 2018.

STAKEHOLDER ENGAGEMENT
for Repeal of Executive Director Second Newborn Screening Rules
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State law requires agencies to establish a representative group of participants when considering to adopt or modify new and existing rules. This is commonly referred to as a stakeholder group.

Early Stakeholder Engagement:
The following individuals and/or entities were invited to provide input and included in the development of these proposed rules:

Organization	**Representative**
Hemoglobinopathies Follow-up Clinic & Colorado Sickle Cell Treatment & Research Center	Donna Holstein, Clinical Educator, Newborn Screening Follow-Up Program Dr. Kathy Hassell, Director and Professor Adult Hematologist
Congenital Hypothyroidism Follow-up Clinic & Rocky Mountain	Dr. Aristides Maniatis, Pediatric Endocrinologist
Congenital Adrenal Hyperplasia Follow-up Clinic	Dr. Jennifer Barker, Pediatric Endocrinologist
Children's Hospital of Colorado/Cystic Fibrosis Follow-up Clinic	Dr. Scott Sagel, Pediatric Pulmonologist
Children's Hospital/Inherited Metabolic Disease Follow-up Clinic	Dr. Peter Baker, Pediatric Metabolic Geneticist Dr. Janet Thomas, Pediatric Metabolic Geneticist Dr. Shawn McCandless, Section Head, Genetics and Metabolism Erica Wright, Senior Instructor, Certified Genetic Counselor
Pediatrician (Western Slope Region)	Dr. Patrice Whistler, Pediatrician
Neonatologist (Colorado Springs)	Dr. Bob Kiley, Neonatologist
Colorado Hospital Association	Amber Burkhardt, Manager, Public Policy
Rocky Mountain Chapter of Cure SMA	Michelle Pritekel
Colorado Midwives Association	Melissa Sexton, Interim President
Children's Hospital of Colorado	Ellen Stern, Senior Policy Coordinator, Government Affairs
Laboratory Services Division staff	Dr. Emily Travanty, Scientific Director Olga Ivanova, Fiscal Services Manager Greg Bonn, Newborn Screening Operations Manager Dr. Sudhindra Rao, Newborn

	Screening Scientist Kyle Senger, Newborn Screening Laboratory Technician Kay Reilly, Newborn Screening Laboratory Technician
Mother of Child with MPS-1	Christine Tippett
Wyoming DoH	Christina Taylor, Women and Infant Health Program Manager Maternal and Child Health Unit
Mother of Child with MCADD	Kay Kelly
Biogen	Ritchard Engelhardt, State Government Affairs
Novartis	Barbara Boner, Director, State Government Affairs
Children's Hospital/Severe Combined Immunodeficiency Follow-up Clinic	Dr. Cullen Dutmer, Pediatric Immunologist Dr. Elena Hsieh, Pediatric Immunologist
Patient	Lori Wise
University of Colorado Hospital (UCSH)	Dr. Mary Kohn, Associate Professor of Clinical Pediatrics
NewSTEPs	Dr. Yvonne Keller-Guenther, Associate Director for NewSTEPs 360 Dr. Marci Sontag, Associate Director for NewSTEPs 360
Parent of Child with Spinal Muscular Atrophy	Nicole Shaklee
Colorado Department of Public Health and Environment	Dr. Tista Ghosh, Interim Chief Medical Officer Margaret Ruttenber, Program Director, Colorado Responds to Children with Special Needs (CRCSN) Rachel Hutson, Branch Chief, Prevention Services Division
March of Dimes	Lyn Elliott, Regional Director, Advocacy & Government Affairs
Colorado Chapter of the American Academy of Pediatrics	Dr. Edward Maynard, Vice President
Pediatrics Section of Children's Hospital of Colorado	Dr. Stephen R. Daniels, Section Head
Rocky Mountain Hospital for Children	Dr. Reginald Washington, Chief Medical Officer
Colorado Children's Campaign	Erin Miller, Vice President, Health Initiatives

A variety of early stakeholder engagements were conducted. These events include activities tied to contract monitoring, as well as community outreach events such as a series of peer-to-peer networking events in August 2018. The Department also met with newborn screening stakeholders on September 25, 2018. Feedback from individual contracted specialists was also

sought to ensure the standards established in the rule could be implemented if the Department sought for the services to be provided by a contractor. Written feedback from Children's Hospital of Colorado and the Colorado Midwives Association is incorporated into the proposed rules.

The Department has continued to engage stakeholders. The Department met with stakeholders in October and November. All feedback has been reviewed and incorporated into the packet when deemed appropriate.

Stakeholder Group Notification
The stakeholder group was provided notice of the rulemaking hearing and provided a copy of the proposed rules or the internet location where the rules may be viewed. Notice was provided prior to the date the notice of rulemaking was published in the Colorado Register (typically, the 10th of the month following the Request for Rulemaking).

____ Not applicable. This is a Request for Rulemaking Packet. Notification will occur if the Board of Health sets this matter for rulemaking.

_X__ Yes.

Summarize Major Factual and Policy Issues Encountered and the Stakeholder Feedback Received. If there is a lack of consensus regarding the proposed rule, please also identify the Department's efforts to address stakeholder feedback or why the Department was unable to accommodate the request.

There were two stakeholder requests that were studied and not incorporated into the rule:

For Section 4.2.5 (Board of Health rulemaking), some stakeholders requested requirements to monitor true positive rates and false positive rates. The Department concluded this was inappropriate. If the Department contracts for follow-up services, the contract would define the services and what would be monitored. As such monitoring will be based on the negotiated scope of work. Further elaborating upon monitoring in rule limits the Department's ability to negotiate its contracts and may interfere with the Department's ability to manage contractor performance.

As discussed in the Statement of Basis and Purpose the Department declined to recommend Executive Director rules related to quality control and education or Board of Health rules concerning quality control and supervision for second screening.

Please identify the determinants of health or other health equity and environmental justice considerations, values or outcomes related to this rulemaking.

This rulemaking and the education and outreach the Department will perform to ensure named submitters and birthing facilities are informed of the regulatory requirements and resources ensure timely newborn screening. Follow-up services link families to care and bridge the birth to short-term and long-term services and supports. The Department provides follow-up services for hemoglobinopathies, a group of conditions that frequently affect African Americans.

Overall, after considering the benefits, risks and costs, the proposed rule:
Select all that apply.

	Improves behavioral health and mental health; or, reduces substance abuse or suicide risk.	X	Reduces or eliminates health care costs, improves access to health care or the system of care; stabilizes individual participation; or, improves the quality of care for unserved or underserved populations.
	Improves housing, land use, neighborhoods, local infrastructure, community services, built environment, safe physical spaces or transportation.		Reduces occupational hazards; improves an individual's ability to secure or maintain employment; or, increases stability in an employer's workforce.
	Improves access to food and healthy food options.		Reduces exposure to toxins, pollutants, contaminants or hazardous substances; or ensures the safe application of radioactive material or chemicals.
X	Improves access to public and environmental health information; improves the readability of the rule; or, increases the shared understanding of roles and responsibilities, or what occurs under a rule.		Supports community partnerships; community planning efforts; community needs for data to inform decisions; community needs to evaluate the effectiveness of its efforts and outcomes.
	Increases a child's ability to participate in early education and educational opportunities through prevention efforts that increase protective factors and decrease risk factors, or stabilizes individual participation in the opportunity.		Considers the value of different lived experiences and the increased opportunity to be effective when services are culturally responsive.
X	Monitors, diagnoses and investigates health problems, and health or environmental hazards in the community.	X	Ensures a competent public and environmental health workforce or health care workforce.
	Other:_____ _____		Other:_____ _____

DEPARTMENT OF PUBLIC HEALTH AND ENVIRONMENT

Laboratory Services Division

NEWBORN SCREENING AND SECOND NEWBORN SCREENING

5 CCR 1005-4

~~NEWBORN SCREENING REGULATIONS~~

SECTION 1: AUTHORITY AND DEFINITIONS

1.1 THESE RULES AND REGULATIONS ARE ESTABLISHED u~~U~~nder the authority contained in Section~~s 25-4-801 through 25-4-804 and~~ 25-4-1001 et seq,~~through 6 (not including Section 25-4-1004.7)~~ C.R.S. ~~(1998), the following rules and regulations are established.~~

1.2 Definitions: The following terms, whenever used in or referred to in these regulations, shall have the following respective meanings, unless a different meaning clearly appears from the context:

"BIRTHING FACILITY" MEANS A GENERAL HOSPITAL OR BIRTHING CENTER LICENSED OR CERTIFIED PURSUANT TO SECTION 25-1.5-103.

“Department” shall mean the Colorado Department of Public Health and Environment.

“EXCEPTIONAL CIRCUMSTANCES” SHALL MEAN CIRCUMSTANCES WITHIN 364 DAYS AFTER THE BIRTH OF THE CHILD, WHERE THE DEPARTMENT, AT ITS SOLE DISCRETION, MAY DETERMINE THAT TIMELY COLLECTION OF A SPECIMEN WAS NOT FEASIBLE, BUT SCREENING REMAINS APPROPRIATE. THIS INCLUDES BUT IS NOT LIMITED TO OBTAINING SPECIMENS FOR CHILDREN BORN OUTSIDE THE UNITED STATES WHO RELOCATE TO COLORADO THROUGH THE ADOPTION PROCESS OR A REFUGEE RESETTLEMENT PROGRAM.

“FOLLOW-UP SERVICES” SHALL MEAN 1) REPEAT OR CONFIRMATORY TESTING IF CLINICALLY NECESSARY AS DETERMINED BY THE DEPARTMENT, OR 2) FOR NEWBORNS THAT SCREEN POSITIVE INITIALLY OR THROUGH REPEAT OR CONFIRMATORY TESTING, REFERRAL SERVICES TO CONNECT NEWBORNS TO THE HEALTHCARE SYSTEM FOR THE PURPOSE OF RECEIVING A DIAGNOSIS, INTERVENTIONS, OR SPECIALTY CARE, AS DETERMINED BY THE DEPARTMENT. FOLLOW-UP SERVICES SUPPORTED OR PERFORMED BY THE DEPARTMENT ARE INTENDED TO FACILITATE RAPID CONNECTION OF NEWBORNS TO APPROPRIATE CARE, BUT ARE NOT INTENDED TO SERVE AS CLINICAL CASE MANAGEMENT SERVICES.

“Initial ~~N~~newborn ~~S~~screening ~~S~~specimen” shall mean, ABSENT EXCEPTIONAL CIRCUMSTANCES, A specimen collected from a newborn~~, prior to discharge~~, BETWEEN 24 AND 48 HOURS AFTER BIRTH AND, TO THE EXTENT FEASIBLE, PRIOR TO ANY BLOOD TRANSFUSION ~~but in all cases within 48 hours after birth for the purpose of conducting screening tests~~.

"Laboratory" shall mean the Colorado Department of Public Health and Environment Laboratory.

"NAMED SUBMITTER" SHALL MEAN THE ENTITY OR INDIVIDUAL IDENTIFIED ON THE DEMOGRAPHIC SLIP ATTACHED TO THE BLOOD SPOT CARD AS THE SUBMITTER OF THAT SPECIMEN.

"NEWBORN" SHALL MEAN A CHILD UNDER 28 DAYS OF AGE WHOSE PARENT(S) OR LEGAL GUARDIAN(S) HAVE NOT OPTED OUT OF NEWBORN BLOOD SPOT SCREENING. NEWBORNS MAY BE REFERRED TO AS "NEONATES."

"SCREEN NEGATIVE" SHALL MEAN A RESULT FROM A SCREENING TEST THAT DOES NOT INDICATE THE PRESENCE OF THE SCREENED CONDITION.

"SCREEN POSITIVE" SHALL MEAN A RESULT FROM A SCREENING TEST THAT INDICATES SOME LIKELIHOOD OF THE SCREENED CONDITION(S) BEING PRESENT, AND THEREFORE REQUIRES FURTHER INVESTIGATION OR TESTING OF THE NEWBORN.

"SECOND NEWBORN SCREENING SPECIMEN" SHALL MEAN A SPECIMEN COLLECTED FROM A NEWBORN BETWEEN 8 AND 14 DAYS AFTER BIRTH FOR THE PURPOSE OF CONDUCTING SECOND SCREENING TESTS.

"SPECIMEN" SHALL MEAN ANY DRIED BLOOD SPOTS COLLECTED, DRIED, AND SUBMITTED TO THE LABORATORY FOR SCREENING.

"TIME-CRITICAL CONDITION" SHALL MEAN A CONDITION IDENTIFIED BY THE DEPARTMENT THAT MAY PRESENT WITH ACUTE SYMPTOMS WITHIN THE FIRST WEEK OF LIFE THEREBY REQUIRING IMMEDIATE TREATMENT TO REDUCE RISK OF DEATH OR INTELLECTUAL OR OTHER PERMANENT DISABILITIES.

"TIME-CRITICAL SCREEN POSITIVE RESULT" SHALL MEAN A POSITIVE SCREENING RESULT THAT SUGGESTS A HIGH LIKELIHOOD OF A TIME-CRITICAL CONDITION.

"TIME-SENSITIVE CONDITION" SHALL MEAN A CONDITION IDENTIFIED BY THE DEPARTMENT NOT TO BE ASSOCIATED WITH EARLY ONSET OF SEVERE SYMPTOMS INCLUDING DEATH OR INTELLECTUAL OR OTHER PERMANENT DISABILITIES.

"TIME-SENSITIVE SCREEN POSITIVE RESULT" SHALL MEAN AN INITIAL NEWBORN SCREENING SPECIMEN RESULT ASSOCIATED WITH ANY RISK LEVEL FOR A TIME-SENSITIVE CONDITION OR A MODERATE RISK LEVEL FOR A TIME-CRITICAL CONDITION, THEREBY ALLOWING TIME FOR COLLECTION AND TESTING OF A SECOND NEWBORN SCREENING SPECIMEN.

"UNSATISFACTORY SPECIMEN" SHALL MEAN A SPECIMEN FOR WHICH ALL TIERS OF TESTING PERFORMED WITHIN THE LABORATORY COULD NOT BE COMPLETED FOR ANY REASON, SUCH AS THE QUALITY OF THE SPECIMEN OR THE AMOUNT OF SPECIMEN PROVIDED.

~~1.3 Procedures~~

SECTION 2: NEWBORN SCREENING REQUIREMENTS FOR NAMED SUBMITTERS

2.1. HYGIENIC COLLECTION CONDITIONS

WORK AREAS USED TO COLLECT SPECIMENS WILL BE CLEAN AND SANITARY. INDIVIDUALS COLLECTING SPECIMENS WILL FOLLOW HYGIENIC PRACTICES INCLUDING HANDWASHING.

2.2 SPECIMEN COLLECTION, HANDLING, AND SUBMISSION

~~1.3.1~~ 2.2.1 Births in ~~Institutions~~ BIRTHING FACILITIES : The blood specimens of newborns born in ~~institutions~~ BIRTHING FACILITIES and all other specimens taken in conformity with the law and these regulations will be sent to the Laboratory for testing. PURSUANT TO SECTION 25-4-1004(1)(B), C.R.S., THE BIRTHING FACILITY WHERE THE INFANT IS BORN SHALL FORWARD ALL SPECIMENS TO THE LABORATORY.

~~Follow up specimens from newborns with positive screening tests will be obtained and tested~~ PURSUANT TO SECTION 25-4-1004(2), C.R.S., THE BIRTHING FACILITY WHERE THE NEWBORN IS BORN SHALL ALSO BE RESPONSIBLE FOR HELPING TO CONNECT INFANTS WHO SCREEN POSITIVE TO FOLLOW-UP SERVICES TO INCLUDE AIDING IN THE COLLECTION OF ADDITIONAL SPECIMENS FOR UNSATISFACTORY SPECIMENS OR SPECIMENS WITH EQUIVOCAL RESULTS, AS WELL AS COLLECTION OF ADDITIONAL SPECIMENS FOR RESOLUTION OF TIME-SENSITIVE AND TIME-CRITICAL SCREEN POSITIVE RESULTS, as necessary for proper diagnosis.

~~1.3.1.1~~ 2.2.1.1 The ~~hospital or institution or the chief medical staff officer or other person in charge thereof~~ BIRTHING FACILITY will ~~cause~~ OBTAIN an initial newborn screening specimen ~~to be obtained~~ from every newborn born therein ~~as late as possible before discharge, but no later than 48 hours of age~~.

~~1.3.1.2~~ 2.2.1.2 The INITIAL NEWBORN SCREENING specimen shall consist of capillary blood collected by heel puncture or alternate method authorized by the Laboratory, PLACED directly ~~up~~on special blotter paper furnished by the Laboratory.

THE INITIAL NEWBORN SCREENING SPECIMEN SHALL BE COLLECTED FROM ALL NEWBORNS AT 24 HOURS OF AGE, BUT NO LATER THAN 48 HOURS OF AGE AND ALWAYS BEFORE THE NEWBORN IS DISCHARGED FROM THE BIRTHING FACILITY, UNLESS EXCEPTIONAL CIRCUMSTANCES EXIST.

HEEL PUNCTURE SAMPLING WILL OCCUR IN A MANNER THAT MAINTAINS THE HEALTH AND SAFETY OF THE NEWBORN AND INDIVIDUAL COLLECTING THE SPECIMEN; ENSURE PROPER LABELING AND PREPARATION OF THE

SPECIMEN FOR DELIVERY, AND ALLOW FOR ACCURATE TEST RESULTS AND PROPER DIAGNOSIS.

All circles shall be saturated with blood from one side of the blotter only. The ~~specimen~~ NAMED submitter will provide, on the attached ~~form~~ DEMOGRAPHIC SLIP, all information requested by the Laboratory.

The specimens~~,~~ SHALL BE ~~after~~ air DRIED HORIZONTALLY FOR THREE TO FOUR HOURS. AFTER AIR-drying, SPECIMENS ~~will~~ SHALL be forwarded to the Laboratory within 24 hours of collection, by courier or overnight delivery if available. SPECIMENS SHALL BE SUBMITTED TO THE LABORATORY IN THE FORM AND MANNER REQUIRED BY THE DEPARTMENT.

~~1.3.1.3~~ 2.2.1.3 If the newborn is to receive a blood transfusion, then the specimen for newborn screening is to be obtained prior to this procedure. IF AN INITIAL NEWBORN SCREENING SPECIMEN IS COLLECTED AFTER TRANSFUSION, THE COLLECTION FORM WILL BE MARKED APPROPRIATELY TO INDICATE TRANSFUSION OCCURRED.

~~1.3.2~~ 2.2.2 Births Outside ~~Institutions~~ BIRTHING FACILITIES: The physician, ~~nurse~~ REGISTERED midwife, or other health professional attending a birth outside a ~~hospital~~ BIRTHING FACILITY, shall be responsible for the collection~~s~~ and forwarding of the specimen described in ~~1.3.1.2~~ 2.2.1.2 ~~above~~. In the absence of a health professional, any other person attending the birth, or in the absence of any person so attending, the PARENT(S) ~~father~~ or ~~mother~~ LEGAL GUARDIAN(S) OF THE NEWBORN, or in the absence of OR INABILITY OF the NEWBORN'S ~~father and the inability of the NEWBORN'S mother~~ PARENT(S) OR LEGAL GUARDIAN(S), the person in charge of the premises where the birth occurred shall be responsible.

~~1.4 Testing and Reporting: The prescribed tests will be initiated by the Laboratory within 24 hours of receipt of the specimen, weekends and holidays excepted. The Laboratory shall report as follows:~~

~~1.4.1 Reports of normal test results will be sent to the submitting agency within seven working days.~~

~~1.4.2 Abnormal test results will be reported immediately by telephone to the physician of record and to designated consultants. In case of inability to identify or locate a physician of record, the abnormal test result will be reported to the hospital or submitting agency which originated the specimen, or, if the birth did not occur in a health facility, to the father or mother.~~

~~1.4.3 Unsatisfactory specimens or specimens with equivocal results will be reported immediately to the submitting agency which originated the specimen with an explanation of the results. The submitting agency responsible for the~~

~~newborn's care at the time of the report will cause another specimen to be forwarded at the appropriate time.~~

~~1.4.4~~ 2.3. CARE COORDINATION

The ~~submitting agency that originated the~~ NAMED SUBMITTER OF AN INITIAL NEWBORN SCREENING specimen shall forward ~~the~~ ANY ~~N~~newborn ~~S~~screening SCREEN NEGATIVE OR SCREEN POSITIVE results PRODUCED BY THE LABORATORY PURSUANT TO RULE 4 to the health care provider responsible for the newborn's care within ~~the time frame of 1.4.1 and 1.4.3 above~~ SEVEN DAYS FOR ANY SCREEN NEGATIVE RESULTS, WITHIN 72 HOURS FOR ANY TIME-SENSITIVE SCREEN POSITIVE RESULTS AND WITHIN 24 HOURS FOR ANY TIME-CRITICAL SCREEN POSITIVE RESULTS.

~~1.5 Quality Control and Education~~

~~1.5.1 The Laboratory shall have available for review a written quality assurance program plan covering all aspects of laboratory activity.~~

~~1.5.2 The Laboratory shall make available educational materials and training concerning specimen collection to all submitting agencies.~~

~~1.6~~2.4 List of Conditions for Newborn Screening

~~1.6.1~~ The Laboratory shall conduct screening tests for the following conditions:

~~1.6.1.1~~ 2.4.1 Phenylketonuria

~~1.6.1.2~~ 2.4.2 Congenital Hypothyroidism

~~1.6.1.3~~ 2.4.3 Hemoglobinopathies

~~1.6.1.4~~ 2.4.4 Galactosemia

~~1.6.1.5~~ 2.4.5 Cystic Fibrosis

~~1.6.1.6~~ 2.4.6 Biotinidase Deficiency

~~1.6.1.7~~ 2.4.7 Congenital Adrenal Hyperplasia

~~1.6.1.8~~ 2.4.8 Medium Chain Acyl-CoA dehydrogenase deficiency

~~1.6.1.9~~ 2.4.9 Very Long Chain Acyl-CoA dehydrogenase deficiency

~~1.6.1.10~~ 2.4.10 Long-Chain L-3-Hydroxy Acyl-CoA dehydrogenase deficiency

~~1.6.1.11~~ 2.4.11 Trifunctional protein deficiency

~~1.6.1.12~~ 2.4.12 Carnitine Acyl-carnitine translocase deficiency

~~1.6.1.13~~ 2.4.13	Short Chain Acyl-CoA dehydrogenase deficiency
~~1.6.1.14~~ 2.4.14	Carnitine palmitoyltransferase II deficiency
~~1.6.1.15~~ 2.4.15	Glutaric acidemia Type 2
~~1.6.1.16~~2.4.16	Arginosuccinic acidemia
~~1.6.1.17~~ 2.4.17	Citrullinemia
~~1.6.1.18~~ 2.4.18	Tyrosinemia
~~1.6.1.19~~ 2.5.19	Hypermethionemia
~~1.6.1.20~~ 2.4.20	Maple Syrup urine disease
~~1.6.1.21~~ 2.4.21	Homocystinuria
~~1.6.1.22~~ 2.4.22	Isovaleric acidemia
~~1.6.1.23~~ 2.4.23	Glutaric acidemia Type 1
~~1.6.1.24~~ 2.5.24	3-hydroxy-3-methylglutaryl-CoA Lyase deficiency
~~1.6.1.25~~ 2.4.25	Multiple Carboxylase deficiency
~~1.6.1.26~~ 2.4.26	3-methylcrotonyl-CoA carboxylase deficiency
~~1.6.1.27~~ 2.4.27	3-methylglutaconic aciduria
~~1.6.1.28~~ 2.4.28	Methylmalonic acidemias
~~1.6.1.29~~ 2.4.29	Propionic acidemia
~~1.6.1.30~~ 2.4.30	beta-Ketothiolase deficiency
~~1.6.1.31~~ 2.4.31	Carnitine uptake defect
~~1.6.1.32~~ 2.4.32	Arginase deficiency
~~1.6.1.33~~ 2.4.33	Malonic acidemia
~~1.6.1.34~~ 2.4.34	Carnitine palmitoyltransferase deficiency 1A
~~1.6.1.35~~ 2.4.35	Severe Combined Immunodeficiency

~~**RULES AND REGULATIONS OF THE EXECUTIVE DIRECTOR COLORADO DEPARTMENT OF PUBLIC HEALTH AND ENVIRONMENT**~~
~~**IMPLEMENTATION OF SECOND NEWBORN SCREENING**~~

~~1.1 Under the authority contained in Section 25-4-1004.5(3) C.R.S., the following Rules and Regulations are established.~~

~~1.2 Definitions~~

~~"Department" shall mean the Colorado Department of Public Health and Environment.~~

~~"Executive Director" shall mean the executive director of the Colorado Department of Public Health and Environment.~~

~~"Laboratory" shall mean the Colorado Department of Public Health and Environment Laboratory.~~

~~"Initial newborn screening specimen" shall mean specimen collected from a newborn prior to discharge, but in all cases within 48 hours after birth for the purpose of conducting screening tests.~~

~~"Second newborn screening specimen" shall mean a specimen collected from a newborn between eight and 14 days after birth, but in no case less than 72 hours or greater than 30 days after birth, for the purpose of conducting screening tests.~~

SECTION 3: SECOND NEWBORN SCREENING REQUIREMENTS FOR NAMED SUBMITTERS

3.1. HYGIENIC COLLECTION CONDITIONS

WORK AREAS USED TO COLLECT SECOND NEWBORN SCREENING SPECIMENS WILL BE CLEAN AND SANITARY. INDIVIDUALS COLLECTING SECOND NEWBORN SCREENING SPECIMENS WILL FOLLOW HYGIENIC PRACTICES INCLUDING HANDWASHING.

3.2 NOTIFICATION, SPECIMEN COLLECTION, HANDLING AND SUBMISSION

3.2.1 NOTIFICATION

~~1.3 Procedures~~

~~1.3.1~~ The parent(s) or other legal guardian(s) of the newborn shall be advised ~~of the necessity of the~~ THAT A second newborn screening test IS REQUIRED FOR CONDITIONS AS SPECIFIED IN RULE 3.2.2.2 AND 3.3.

~~1.3.1.1~~ 3.2.1.1 Births in ~~Institutions~~ BIRTHING FACILITIES: It shall be the responsibility of the ~~hospital or institution or the chief medical staff officer or other person in charge thereof~~ BIRTHING FACILITY to advise, verbally and in writing, such as by written information made available from the Department, the parent(s) or other legal guardian(s) of the newborn that it is necessary to have a second newborn screening test performed.

~~1.3.1.2~~ 3.2.1.2 Births outside ~~Institutions~~ BIRTHING FACILITIES: It shall be the responsibility of the physician, ~~nurse midwife,~~ ~~lay~~ REGISTERED midwife, or other health professional attending a birth outside a ~~hospital~~ BIRTHING FACILITY to advise, verbally and in writing, such as by written information made available from the Department, the parent(s) or other legal guardian(s) of the newborn~~, of the necessity of the~~ THAT IT IS NECESSARY TO HAVE A second newborn screening PERFORMED.

3.2.2 COLLECTION

~~1.3.2~~ 3.2.2.1 The attending health care provider shall collect or require the specimen be collected from all newborns at ~~the first post partum~~ A NEWBORN WELL CHILD appointment BETWEEN 8 AND 14 DAYS AFTER BIRTH~~, but in no case less than 72 hours or greater than 30 days after birth~~.

The specimen shall consist of capillary blood collected by heel puncture or alternate method authorized by the Laboratory, PLACED directly ~~up~~on special blotter paper furnished by the Laboratory.

HEEL PUNCTURE SAMPLING WILL OCCUR IN A MANNER THAT MAINTAINS THE HEALTH AND SAFETY OF THE NEWBORN AND INDIVIDUAL COLLECTING THE SPECIMEN; ENSURE PROPER LABELING AND PREPARATION OF THE SPECIMEN FOR DELIVERY, AND ALLOW FOR ACCURATE TEST RESULTS AND PROPER DIAGNOSIS.

All circles shall be saturated with blood from one side of the blotter only. The NAMED submitter will provide, on the attached ~~form~~ DEMOGRAPHIC SLIP, all information requested by the Laboratory.

The specimens~~, after~~ SHALL BE air ~~drying~~ DRIED HORIZONTALLY FOR THREE TO FOUR HOURS. ~~The specimens, a~~After air-drying, SPECIMENS shall be forwarded to the Laboratory within 24 hours of collection by first class mail, courier, or overnight delivery. SPECIMENS SHALL BE SUBMITTED TO THE LABORATORY IN THE FORM AND MANNER REQUIRED BY THE DEPARTMENT.

~~1.4 Testing and Reporting: The prescribed tests will be initiated by the Laboratory within 24 hours of receipt of the specimen, weekends and holidays excepted. The Laboratory shall report as follows:~~

~~1.4.1 Reports of normal test results will be sent to the submitting agency within seven working days.~~

~~1.4.2 Abnormal test results will be reported immediately by telephone to the physician of record and to designated consultants. In case of inability to identify or locate a physician of record, the abnormal test result will be reported to the submitting agency which originated the specimen, or, if the birth did not occur in a health facility, to the father or mother.~~

~~1.4.3 Unsatisfactory specimens or specimens with equivocal results will be reported immediately to the submitting agency which originated the specimen with an explanation of the results. The health care provider responsible for the newborn's care at the time of the report will cause another specimen to be forwarded at the appropriate time.~~

~~1.4.4 The submitting agency that originated the specimen shall forward the newborn screening results to the health care provider responsible for the newborn's care.~~

~~1.5 Quality Control and Education~~

~~1.5.1 The Laboratory shall have available for review a written quality assurance program plan covering all aspects of testing and reporting second NEWBORN SCREENING specimens.~~

~~1.5.2 The Laboratory shall make available educational materials and training concerning SECOND NEWBORN SCREENING specimen collection to submitting agencies.~~

3.2.2.2 SECTION 25-4-1004.5(3)(b)(V), C.R.S., ALLOWS EXCEPTIONS TO TESTING OF SECOND NEWBORN SCREENING SPECIMENS. SECOND NEWBORN SCREENING SPECIMEN TESTING IS NOT REQUIRED FOR THE CONDITIONS IDENTIFIED AT 3.3.4, 3.3.5 AND 3.3.6 UNLESS: AN UNSATISFACTORY SPECIMEN WAS SUBMITTED FOR AN INITIAL NEWBORN SCREENING SPECIMEN; AN ABNORMAL RESULT WAS OBTAINED ON AN INITIAL NEWBORN SCREENING SPECIMEN FROM THE SAME NEWBORN, OR; THERE IS NO RECORD OF A SATISFACTORY INITIAL NEWBORN SCREENING SPECIMEN SUBMISSION.

~~1.6~~ 3.3 List of Conditions for Second Newborn Screening

~~1.6.1~~ The Laboratory shall conduct screening tests for the following conditions:

~~1.6.1.1~~ 3.3.1 Phenylketonuria

~~1.6.1.2~~ 3.3.2 Congenital Hypothyroidism

~~1.6.1.3~~ 3.3.3 Hemoglobinopathies

~~1.6.1.4~~ 3.3.4 Galactosemia ~~1~~

~~1.6.1.5~~ 3.3.5 Cystic Fibrosis ~~1~~

~~1.6.1.6~~ 3.3.6 Biotinidase Deficiency ~~1~~

~~1.6.1.7~~ 3.3.7 Congenital Adrenal Hyperplasia

1) ~~These disorders need not be tested again unless:~~

a) ~~an unsatisfactory specimen was submitted for the first screen testing, or~~
b) ~~an abnormal result was obtained on the first screen testing, or~~
c) ~~no record of a satisfactory first screen specimen submission can be ascertained.~~

SECTION 4: LABORATORY TESTING, REPORTING AND FOLLOW-UP SERVICES FOR NEWBORN SCREENING AND SECOND NEWBORN SCREENING

4.1 THE LABORATORY SHALL OPERATE AT LEAST SIX (6) DAYS PER WEEK. SPECIMEN TESTING WILL BE INITIATED BY THE LABORATORY ON THE DATE OF RECEIPT OR THE NEXT OPERATING DAY FOLLOWING RECEIPT OF THE SPECIMEN.

RESULTS WILL BE SENT TO THE NAMED SUBMITTER FOR INITIAL NEWBORN SCREENING AND FOR SECOND NEWBORN SCREENING. RESULTS WILL BE REPORTED IN A MANNER AND ON A TIMELINE CONSISTENT WITH THE URGENCY OF INTERVENTION.

4.1.1 REPORTS OF SCREEN NEGATIVE TEST RESULTS WILL BE SENT WITHIN SEVEN WORKING DAYS.

4.1.2 AN ATTEMPT TO REPORT TIME-CRITICAL SCREEN POSITIVE RESULTS WILL BE MADE IMMEDIATELY, BUT IN NO CASE LONGER THAN 24 HOURS. REPORTING MAY OCCUR THROUGH THE DEPARTMENT OR ITS CONTRACTOR. ATTEMPTS TO REPORT TIME-CRITICAL SCREEN POSITIVE RESULTS WILL CONTINUE FOR UP TO 180 DAYS.

4.1.2.1 IF A CONTRACTOR IS UTILIZED BY THE DEPARTMENT, THE CONTRACTOR MAY RECEIVE IDENTIFYING PATIENT INFORMATION, PROTECTED HEALTH INFORMATION, NAMED SUBMITTER INFORMATION AND ATTENDING HEALTH CARE PROVIDER INFORMATION TO THE EXTENT NECESSARY TO PERFORM THESE DUTIES AND IN THE MANNER AUTHORIZED BY LAW.

4.1.3 AN ATTEMPT TO REPORT TIME-SENSITIVE SCREEN POSITIVE RESULTS WILL BE MADE IMMEDIATELY, BUT IN NO CASE LONGER THAN 72 HOURS. REPORTING MAY OCCUR THROUGH THE DEPARTMENT OR ITS CONTRACTOR. ATTEMPTS TO REPORT TIME-SENSITIVE SCREEN POSITIVE RESULTS WILL CONTINUE FOR UP TO 180 DAYS.

4.1.3.1 IF A CONTRACTOR IS UTILIZED BY THE DEPARTMENT, THE CONTRACTOR MAY RECEIVE IDENTIFYING PATIENT INFORMATION, PROTECTED HEALTH INFORMATION, NAMED SUBMITTER INFORMATION AND ATTENDING HEALTH CARE PROVIDER INFORMATION TO THE EXTENT NECESSARY TO PERFORM THESE DUTIES AND IN THE MANNER AUTHORIZED BY LAW.

4.1.4 AN ATTTEMPT TO REPORT UNSATISFACTORY SPECIMENS OR SPECIMENS WITH EQUIVOCAL RESULTS WILL BE MADE IMMEDIATELY, BUT IN NO CASE LONGER THAN 48 HOURS.

4.2 FOLLOW-UP SERVICES

THE FOLLOWING RULES APPLY TO FOLLOW-UP SERVICES, WHILE RECOGNIZING THAT FAMILY PARTICIPATION IN THE FOLLOW-UP SUPPORT AND ASSISTANCE SERVICES IS VOLUNTARY.

4.2.1 TIMEFRAME FOR INITIATING SERVICES

4.2.1.1 FOR TIME-CRITICAL SCREEN POSITIVE RESULTS, FOLLOW-UP SERVICES WILL BE INITIATED WITHIN FOUR HOURS OR THE CLINICALLY-RELEVANT TIMEFRAME AUTHORIZED BY THE DEPARTMENT TO PREVENT DEATH OR INTELLECTUAL OR OTHER PERMANENT DISABILITIES.

4.2.1.2 FOR TIME-SENSITIVE SCREEN POSITIVE RESULTS, FOLLOW-UP SERVICES WILL BE INITIATED WITHIN A CLINICALLY-RELEVANT TIMEFRAME TO PREVENT DEATH OR INTELLECTUAL OR OTHER PERMANENT DISABILITIES. WHEN REQUIRED BY THE DEPARTMENT, FOLLOW-UP SERVICES WILL BEGIN WITH REPEAT OR CONFIRMATORY TESTING.

4.2.2 REPEAT OR CONFIRMATORY TESTING

REPEAT OR CONFIRMATORY TESTING WILL OCCUR WHEN CLINICALLY NECESSARY AS DETERMINED BY THE DEPARTMENT. IF, THROUGH REPEAT OR CONFIRMATORY TESTING, THE NEWBORN SCREENING RESULT IS SCREEN NEGATIVE, FOLLOW-UP SERVICES WILL BE DISCONTINUED AFTER COMMUNICATING THE RESULT.

4.2.3 TIMEFRAME FOR PROVIDING REFERRAL SERVICES

REFERRALS TO SPECIALISTS WILL OCCUR IN A TIMELY MANNER SO A PARENT OR LEGAL GUARDIAN MAY MAKE A TIMELY DECISION TO RESPOND TO A TIME-CRITICAL SCREEN POSITIVE RESULT OR A TIME-SENSITIVE SCREEN POSITIVE RESULT. ALL REFERRALS MUST OCCUR WITHIN THE FIRST 28 DAYS OF AGE UNLESS, AT ITS DISCRETION, THE DEPARTMENT MAY EXTEND FOLLOW-UP SERVICES BEYOND 28 DAYS OF AGE WHEN REPEAT OR CONFIRMATORY TESTING, DIAGNOSIS, INTERVENTIONS HAVE CREATED NECESSARY DELAYS TO THE DEPARTMENT'S ABILITY TO PROVIDE REFERRAL SERVICES OR EXCEPTIONAL CIRCUMSTANCES EXIST. IN NO INSTANCE WILL FOLLOW-UP SERVICES CONTINUE BEYOND 365 DAYS OF THE CHILD'S BIRTH.

4.2.4 IF A CONTRACTOR IS UTILIZED BY THE DEPARTMENT TO PERFORM FOLLOW-UP SERVICES, THE CONTRACTOR MAY RECEIVE IDENTIFYING PATIENT INFORMATION, PROTECTED HEALTH INFORMATION, NAMED SUBMITTER INFORMATION AND ATTENDING

HEALTH CARE PROVIDER INFORMATION TO THE EXTENT NECESSARY TO PERFORM THESE DUTIES AND IN THE MANNER AUTHORIZED BY LAW.

4.2.5 MONITORING PARTICIPATION IN FOLLOW-UP SERVICES

THE DEPARTMENT SHALL MONITOR:

4.2.5.1 THE NUMBER OF NEWBORNS WITH A SCREEN POSITIVE RESULT WHO OPT TO NOT PARTICIPATE IN FOLLOW-UP SERVICES;

4.2.5.2 THE NUMBER OF NEWBORNS WITH A SCREEN POSITIVE RESULT WHO RECEIVE REPEAT AND CONFIRMATORY TESTING WHEN CLINICALLY NECESSARY;

4.2.5.3 THE NUMBER OF NEWBORNS WITH A SCREEN POSITIVE RESULT WHO RECEIVE REFERRAL SERVICES;

4.2.5.4 THE NUMBER OF NEWBORNS WITH A SCREEN POSITIVE RESULT WHO MOVE OUT OF STATE, WITHDRAW FROM OR DO NOT PARTICIPATE IN FOLLOW-UP SERVICES, AND;

4.2.5.5 SUCH OTHER MONITORING THE DEPARTMENT DEEMS APPROPRIATE TO MONITOR THE EFFECTIVENESS OF NEWBORN SCREENING, SECOND NEWBORN SCREENING AND FOLLOW-UP SERVICES.

4.3 IF A CONTRACTOR IS UTILIZED BY THE DEPARTMENT, THE CONTRACTOR MAY RECEIVE IDENTIFYING PATIENT INFORMATION, PROTECTED HEALTH INFORMATION, NAMED SUBMITTER INFORMATION AND ATTENDING HEALTH CARE PROVIDER INFORMATION TO THE EXTENT NECESSARY TO PERFORM THESE DUTIES AND IN THE MANNER AUTHORIZED BY LAW.