COLORADO
Department of Public Health & Environment

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To: Members of the State Board of Health

From: Dr. Darren Michael, Newborn Screening Program Manager, Laboratory Services Division

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

Date: October 18, 2018

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

In preparation for a Public Rulemaking Hearing, please find copies of the following documents:

a) Proposed Amendments to 5 CCR 1005-4,
b) Statement of Basis and Purpose and Specific Statutory Authority,
c) Regulatory Analysis, and
d) Early Stakeholder Engagement

As summarized in the fiscal note for HB 18-1006:

> When an infant is born in Colorado, the two blood samples are collected and forwarded to the CDPHE, which tests for 37 rare genetic and metabolic conditions. The first sample is taken by the hospital, birthing facility, or midwife within 48 hours of birth; the second sample 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.

While the overall set of rules is titled “Newborn Screening and Second Newborn Screening,” there are two sub-sections titled “Newborn Screening Regulations” and “Implementation of Second Newborn Screening.” Together, these definitions, procedures and requirements establish roles and responsibilities, as well as best practices, for the metabolic testing portion of Colorado’s Newborn Screening Program. The majority of the rule is promulgated through

the State Board of Health; however, the Executive Director had rulemaking authority over aspects of second newborn screening.

Recent legislation (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.

Section 25-4-1004(3), C.R.S. states:

> The state board shall promulgate rules concerning the requirements of the newborn screening program for genetic and metabolic disorders, including: (i) in addition to those conditions listed in subsection (1)(b) Of this section, any other conditions for which testing must occur;(ii) obtaining samples or specimens from newborn infants required for the tests prescribed by the state board; and (iii) the handling and delivery of samples or specimens for testing and examination.

Board of Health authority to promulgate rules that identify which conditions will require a second screening is unchanged, Section 25-4-1004.5(3)(a), C.R.S. However, HB 18-1006 transferred the Executive Director's rulemaking authority to establish the standards for second specimen testing to the Board of Health. Section 25-4-1004.5(3)(b), C.R.S. now reads:

> The state board [of health] is authorized to promulgate rules and standards for the implementation of the second specimen testing specified in this subsection (3), including: (I) Identification of those conditions for which a second specimen shall be required; (II) The age of the infant at which the second screening may be administered; (III) The method by which the parent or parents of a newborn shall be advised of the necessity for a second specimen test; (IV) The procedure to be followed in administering the second specimen test; (V) Any exceptions to the necessity for a second specimen test and the procedures to be followed in such cases; and (VI) The standards of supervision and quality control that shall apply to second specimen testing.

Though the rulemaking authority has changed, the rulemaking authorization is substantively the same. To effectuate the change the Department requests the Executive Director repeal the Executive Director rules and that the Board of Health adopt the second newborn screening rules as modified herein.

HB 18-1006 also expanded the Board of Health's rulemaking authority to include follow-up services. Section 25-4-1005.4(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.)

The proposed revisions implement this new 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.

This rulemaking does not add new conditions or modify the list of conditions on the panel.

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

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," "Urgent Screen Positive," "Non-urgent Screen Positive Result," "Time-Critical Condition," and "Time-Sensitive Condition" are taken from or adapted from the cited Clinical and Laboratory Standards Institute document (1) or the cited position statement by the Society for Inherited Metabolic Disorders (4). "Follow up" is also defined as described below in item 4b.

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 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, quality control and information about the programs is required 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.

Section 25-4-1004.5(3), C.R.S., does require the Board of Health to promulgate rules governing supervision and quality control standards for second specimen testing.

3) 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 community, which is the intended audience for this rule. Also, the term 'post partum' was removed to avoid confusion for obstetrics and gynecology physicians, who typically see mothers approximately six weeks after delivery.

4) 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. The Department needs to manage false-positives and thus, the rule recognizes that the department may require repeat or confirmatory testing prior to initiating referral services. The proposed rule then establishes the time frame in which follow-up services may occur.

      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 apply. These programs and their service providers may also be of assistance to these newborns and their families. The statute references medical providers and specialists so at this time the proposed 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 is not included in the definition of follow-up services. Monitoring in the proposed rule 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, monitor information on children diagnosed with conditions in the newborn screen. CRCSN is authorized to conduct population-level health surveillance, including relevant medical record review and examination of risk factors, up to age 3, on cases with these conditions and other birth defects, pursuant to C.R.S. 25-1.5-101 – 25-1.5-105, C.R.S. 25 – 1- 122 and Board of Health Regulations, 6 CCR-1009-7 (State of Colorado Rules and Regulations Pertaining to the Detection, Monitoring, and Investigation of Environmental and Chronic Diseases). Public health monitoring or population health surveillance involves the ongoing collection, analysis, and dissemination of health data to prevent and control disease and injury in a community, as defined by the Centers for Disease Control and Prevention (CDC). In addition to these health surveillance activities, CRCSN also provides parents with information on community resources for their children. It is an unnecessary duplication and expense to extend newborn screening monitoring beyond what is needed to perform effective screening and follow-up services.

To the extent to which the Department performs these services through a contractor, the contract will comport with the Procurement Code, State Fiscal and Personnel rules, and Department policies governing contract monitoring, privacy and data use. As written the proposed rule requires referral services in all screen positive cases as this is directed by the statute.

5) Statutory citations, terminology, clarifying edits and formatting to improve readability are proposed throughout the rule. The terms mother and father and replace them gender neutral terms "parent(s)" and "legal guardian(s)."

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 the Department will use to determine whether it should recommend a condition be added. 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

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

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 impact Birthing Facilities, Midwives, Reference Laboratories (e.g. Quest, LabCorp), Pediatrician's Offices, Family Medicine Offices, Clinical Specialists currently contracted with CDPHE to provide follow-up services (~20), Patient Advocacy Groups (e.g. March of Dimes, Cure SMA, etc.), Adult Patients with Rare Congenital Disorders, Colorado's Newborns (~67,000/yr), and Parents/Families of Colorado's Newborns

   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:

      Birthing Facilities, Midwives, Reference Laboratories (e.g. Quest, LabCorp), Pediatrician's Offices, Family Medicine Offices.

   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:

      Clinical Specialists currently contracted with CDPHE to provide follow-up services (~20), Patient Advocacy Groups (e.g. March of Dimes, Cure SMA, etc.), Adult Patients with Rare Congenital Disorders

   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:

      Colorado's Newborns (~67,000), Parents/Families of Colorado's Newborns

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. Through HB 18-1006, the General Assembly authorized the Department to increase the current fee from $92 to $111.00. The fee adjustment increases the newborn screening portion of the Newborn Genetics Cash Fund by $1.3 million (The legislation also authorized additional funding for newborn hearing screening; however, that is not included in this rule.) We do not envision any increase in costs for stakeholders. The fee increase supports the newborn screening program generally and funds the activities delineated in HB 18-1006.

The fee increase will support the follow-up services provided by the Department or its designee.

Favorable non-economic outcomes:

Timely screening and follow-up services that bridge the birth to pediatric services benefit the medical providers involved. Pediatricians benefit directly from the follow-up services covered by the newborn screening program, as these services provide immediate access to expert medical advice for families and primary care physicians of affected children.

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 captured and why: NA	Dollar amounts that have not been captured and why: NA

Local Government Impact: NA

Statement from HB 18-1006 Fiscal Note:

HB 18-1006 authorized the Department to increase the fee to improve the newborn screening program, including follow-up services. An additional $1.3 million will be generated.

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 designee 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 has reviewed the language for feasibility and policy direction but there are no anticipated cost increases.

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 less wasted activity, e.g. 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, non-urgent screen positive, or urgent screen positive results and ensure timely processing for newborns and their families.

Financial costs to these individuals/entities:

No new costs are expected for these entities and individuals.

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

Pediatricians benefit directly from the follow-up contracts covered by the newborn screening program, as these contracts provide immediate access to expert medical advice for families and primary care physicians of affected children.

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 fiscal note to HB 18-1006 acknowledged that 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 infants 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 will continue. 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
and Amendments to
5 CCR 1005-4, Newborn Screening and Second Newborn Screening

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	Donna Holstein & Dr. Kathy Hassell
Congenital Hypothyroidism Follow-up Clinic	Dr. Aristides Maniatis
Congenital Adrenal Hyperplasia Follow-up Clinic	Dr. Jennifer Barker
Children's Hospital of Colorado/Cystic Fibrosis Follow-up Clinic	Dr. Scott Sagel
Children's Hospital/Inherited Metabolic Disease Follow-up Clinic	Dr. Peter Baker
Children's Hospital/Severe Combined Immunodeficiency Follow-up Clinic	Dr. Elena Hsieh
Pediatrician (Western slope region)	Dr. Patrice Whistler
Neonatologist (Colorado Springs)	Dr. Bob Kiley
Colorado Hospital Association	Amber Burkhardt
Colorado Midwives Association	Melissa Sexton
Children's Hospital of Colorado	Ellen Stern
Laboratory Services Division staff	Dr. Emily Travanty Olga Ivanova Greg Bonn Dr. Sudhindra Rao Kyle Senger
Mother of Child with MPS-1	Christine Tippett
Wyoming DoH	Christina Taylor
Mother of Child with MCADD	Kay Kelly
Children's Hospital/ Inherited Metabolic Disease Follow-up Clinic	Dr. Janet Thomas
Biogen	Ritchard Engelhardt
Novartis	Barbara Boner
Children's Hospital/Severe Combined Immunodeficiency Follow-up Clinic	Dr. Cullen Dutmer
Patient	Lori Wise
University of Colorado Hospital (UCSH)	Dr. Mary Kohn

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 will continue to engage stakeholders. Additional feedback is anticipated. The Department will continue to meet with stakeholders in October and November. All feedback will be reviewed and incorporated into the packet if the Department determines it is appropriate to do so.

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).

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

____ 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.

The Department has modified the rule language based upon its internal review, HB 18-1006 and external feedback received. The Department will continue to engage stakeholders in the coming weeks. If major issues are encountered, the Department is committed to working through them with stakeholders.

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. CDPHE 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

SECTION 1: AUTHORITY AND DEFINITIONS

1.1 THESE RULES AND REGULATIONS ARE ESTABLISHED u~~U~~nder the authority contained in ~~Sections 25-4-801 through 25-4-804~~ and 25-4-1001~~4~~ AND ~~through~~ 25-4-1006~~4~~ ~~(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 BIRTH, WHERE THE DEPARTMENT, AT ITS SOLE DISCRETION, MAY DETERMINE TIMELY COLLECTION OF A SPECIMEN WAS NOT FEASIBLE BUT SCREENING REMAINS APPROPRIATE. THIS INCLUDES BUT IS NOT LIMITED TO OBTAINING SAMPLES 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 DEAPRTMENT, 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, AN INTERVENTION, OR SPECIALTY CARE. FOLLOW-UP SERVICES DOES NOT INCLUDE ANY HEALTHCARE SYSTEM SERVICES.

"Initial ~~N~~newborn ~~S~~screening ~~S~~specimen" shall mean ABSENT EXCEPTIONAL CIRCUMSTANCE, 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 AS THE SUBMITTER OF THE SPECIMEN ON THE DEMOGRAPHIC SLIP ATTACHED TO THE BLOOD SPOT CARD.

"NEWBORN" SHALL MEAN A CHILD UNDER 28 DAYS OF AGES. NEWBORNS MAY BE REFERRED TO AS "NEONATES."

"NON-URGENT 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.

"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 SCREENING TEST THAT INDICATES SOME LIKELIHOOD OF THE SCREENED CONDITION(S) BEING PRESENT, AND FURTHER INVESTIGATION OR TESTING OF THE NEWBORN IS REQUIRED.

"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.

"TIME-CRITICAL CONDITION" SHALL MEAN A CONDITION IDENTIFIED BY THE DEPARTMENT TO BE ASSOCIATED WITH EARLY ONSET OF SEVERE SYMPTOMS INCLUDING DEATH OR INTELLECTUAL OR OTHER PERMANENT DISABILITIES.

"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.

"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.

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

SECTION 2: NEWBORN SCREENING REQUIREMENTS FOR NAMED SUBMITTERS

2.1. HYGIENIC COLLECTION CONDITIONS

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

2.2 SPECIMEN COLLECTION, HANDLING, AND SUBMISSION

2.2.1 Births in BIRTHING FACILITIES ~~Institutions~~: The blood specimens of newborns born in BIRTHING FACILITIES ~~institutions~~ 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 INFANT 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 NON-URGENT AND URGENT SCREEN POSITIVE RESULTS, as necessary for proper diagnosis.

2.2.1.1 The ~~hospital or institution or the chief medical staff officer or other person in charge thereof~~ BIRTHING FACILITY will cause 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~~.

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

THE INITIAL NEWBORN SCREENING SPECIMEN SHALL BE COLLECTED AT 24 HOURS OF AGE, BUT NO LATER THAN 48 HOURS OF AGE.

HEEL PUNCTURE SAMPLING WILL OCCUR IN A MANNER THAT MAINTAINS THE HEALTH AND SAFETY OF THE NEWBORN AND INDIVIDUAL COLLECTING THE SAMPLE; 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 submitter will provide, on the attached form, all information requested by the Laboratory.

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

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 A SPECIMEN IS COLLECTED AFTER TRANSFUSION, THE COLLECTION FORM WILL BE MARKED APPROPRIATELY TO INDICATE TRANSFUSION OCCURRED.

2.2.2 Births Outside BIRTHING FACILITIES ~~Institutions~~: The physician, ~~nurse~~ REGISTERED midwife, or other health professional attending a birth outside a BIRTHING FACILITY ~~hospital~~, shall be responsible for the collections and forwarding of the specimen described in 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), or in the absence of the father and the inability of the mother, 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~~ NAMED SUBMITTER that originated the specimen shall forward the ~~N~~newborn ~~S~~screening SCREEN NEGATIVE AND 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 SCREEN NEGATIVE RESULTS, WITHIN 72 HOURS FOR NON-URGENT SCREEN POSITIVE RESULTS AND WITHIN 24 HOURS FOR URGENT 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.~~

~~6~~2.4 List of Conditions for Newborn Screening

The Laboratory shall conduct screening tests for the following conditions:

2.4.1 Phenylketonuria

2.4.2 Congenital Hypothyroidism

2.4.3 Hemoglobinopathies

2.4.4 Galactosemia

2.4.5 Cystic Fibrosis

2.4.6 Biotinidase Deficiency

2.4.7 Congenital Adrenal Hyperplasia

2.4.8 Medium Chain Acyl-CoA dehydrogenase deficiency

2.4.9 Very Long Chain Acyl-CoA dehydrogenase deficiency

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

2.4.11 Trifunctional protein deficiency

2.4.12 Carnitine Acyl-carnitine translocase deficiency

2.4.13 Short Chain Acyl-CoA dehydrogenase deficiency

2.4.14 Carnitine palmitoyltransferase II deficiency

2.4.15 Glutaric acidemia Type 2

2.4.16 Arginosuccinic academia

2.4.17 Citrullinemia

2.4.18 Tyrosinemia

2.5.19 Hypermethionemia

2.4.20 Maple Syrup urine disease

2.4.21 Homocystinuria

2.4.22 Isovaleric academia

2.4.23 Glutaric acidemia Type 1

2.5.24 3-hydroxy-3-methylglutaryl-CoA Lyase deficiency

2.4.25 Multiple Carboxylase deficiency

2.4.26 3-methylcrotonyl-CoA carboxylase deficiency

2.4.27 3-methylglutaconic aciduria

2.4.28 Methylmalonic acidemias

2.4.29 Propionic academia

2.4.30 beta-Ketothiolase deficiency

2.4.31 Carnitine uptake defect

2.4.32 Arginase deficiency

2.4.33 Malonic academia

2.4.34 Carnitine palmitoyltransferase deficiency 1A

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 SAMPLES WILL BE CLEAN AND SANITARY. INDIVIDUALS COLLECTING SAMPLES WILL FOLLOW HYGIENIC PRACTICES INCLUDING HANDWASHING.

3.2 NOTIFICATION, SPECIMEN COLLECTION, HANDLING AND DELIVERY

3.2.1 NOTIFICATION

~~1.3 Procedures~~

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 IDENTIFIED IN RULE 3.3.

3.2.1 Births in BIRTHING FACILITIES ~~Institutions~~: 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.

3.2.2 Births outside BIRTHING FACILITIES ~~Institutions~~: It shall be the responsibility of the physician, ~~nurse midwife, lay~~ REGISTERED

midwife, or other health professional attending a birth outside a BIRTHING FACILITY ~~hospital~~ 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 second newborn screening.

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, directly upon 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 SAMPLE; 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 submitter will provide, on the attached form, all information requested by the Laboratory.

The specimens~~, after drying~~ WILL BE air DRIED FOR THREE TO FOUR HOURS. ~~The~~ Sspecimens, after air drying, shall be forwarded to the Laboratory within 24 hours of collection by first class mail, courier, or overnight delivery. SPECIMENS WILL BE SUBMITTED 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 specimens.~~

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

3.2.2.2 SECTION 25-4-1004.5(3)(b)(IV), C.R.S., ALLOWS EXCEPTIONS TO SECOND SCREENING SPECIMEN COLLECTION. SECOND SCREENING SPECIMEN COLLECTION 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 FIRST SCREEN TESTING; AN ABNORMAL RESULT WAS OBTAINED ON FIRST SCREEN TESTING, OR; THERE IS NO RECORD OF A SATISFACTORY FIRST SCREEN SPECIMEN SUBMISSION.

3.3 List of Conditions for Second Newborn Screening

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

3.3.1 Phenylketonuria

3.3.2 Congenital Hypothyroidism

3.3.3 Hemoglobinopathies

3.3.4 Galactosemia ~~1~~

3.3.5 Cystic Fibrosis ~~1~~

3.3.6 Biotinidase Deficiency ~~1~~

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 TESTS WILL BE INITIATED BY THE LABORATORY IN THE NEXT DAILY BATCH OF SPECIMEN PROCESSING AFTER RECEIPT OF THE SPECIMEN WITH THE LABORATORY OPERATING AT LEAST SIX (6) DAYS PER WEEK.

RESULTS WILL BE SENT TO THE NAMED SUBMITTER FOR 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 URGENT RESULTS WILL BE MADE IMMEDIATELY, BUT IN NO CASE LONGER THAN 24 HOURS. REPORTING MAY OCCUR THROUGH THE DEPARTMENT OR ITS DESIGNEE. ATTEMPTS TO REPORT URGENT RESULTS WILL CONTINUE FOR UP TO SIX MONTHS.

4.1.2.1 IF A DESIGNEE IS UTILIZED BY THE DEPARTMENT, THE DESIGNEE 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 NON-URGENT RESULTS WILL BE MADE IMMEDIATELY, BUT IN NO CASE LONGER THAN 72 HOURS. REPORTING MAY OCCUR THROUGH THE DEPARTMENT OR ITS DESIGNEE. ATTEMPTS TO REPORT NON-URGENT RESULTS WILL CONTINUE FOR UP TO SIX MONTHS.

4.1.3.1 IF A DESIGNEE IS UTILIZED BY THE DEPARTMENT, THE DESIGNEE 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 URGENT POSITIVE SCREEN 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 NON-URGENT POSITIVE SCREEN 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.

4.2.1 REPEAT OR CONFIRMATORY TESTING

REPEAT OR CONFIRMATORY TESTING WILL OCCUR WHEN CLINICALLY NECESSARY AS DETERMINED BY THE DEAPRTMENT. 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 WITHIN THE FIRST 27 DAYS OF LIFE. AT ITS DISCRETION, THE DEPARTMENT MAY EXTEND FOLLOW-UP SERVICES BEYOND 27 DAYS OF LIFE WHEN REPEAT OR CONFIRMATORY TESTING, DIAGNOSIS, INTERVENTIONS HAVE CREATED NECESSARY DELAYS TO THE DEPARTMENT'S ABILITY TO PROVIDE REFERRAL SERVICES. IN NO INSTANCE WILL FOLLOW-UP SERVICES CONTINUE BEYOND 180 DAYS OF THE CHILD'S BIRTH.

4.2.4 IF A DESIGNEE IS UTILIZED BY THE DEPARTMENT TO PERFORM FOLLOW-UP SERVICES, THE DESIGNEE 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 POSITIVE SCREEN RESULT WHO OPT TO NOT PARTICIPATE IN FOLLOW-UP SERVICES;

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

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

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

4.3 IF A DESIGNEE IS UTILIZED BY THE DEPARTMENT, THE DESIGNEE 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.