DEPARTMENT OF PUBLIC HEALTH AND ENVIRONMENT

Laboratory Services Division

NEWBORN SCREENING AND SECOND NEWBORN SCREENING

5 CCR 1005-4

[Editor's Notes follow the text of the rules at the end of this CCR Document.]

Adopted by the Board of Health on March 17, 2021. Effective May 15, 2021.

SECTION 1: AUTHORITY AND DEFINITIONS

1.1 These rules and regulations are established under the authority contained in Section 25-4-1001 et seq., C.R.S.

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, C.R.S.

"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 newborn screening specimen" shall mean, absent exceptional circumstances, a specimen collected from a newborn, between 24 and 48 hours after birth and, to the extent feasible, prior to any blood transfusion.

"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.

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

2.2.1 Births in birthing facilities: the blood specimens of newborns born in 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.

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.

2.2.1.1 The birthing facility will obtain an initial newborn screening specimen from every newborn born therein.

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 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 named submitter will provide, on the attached demographic slip, all information requested by the Laboratory.

The specimens shall be air dried horizontally for three to four hours. After air-drying, specimens 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.

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.

2.2.2 Births outside birthing facilities: the physician, registered midwife, or other health professional attending a birth outside a birthing facility, shall be responsible for the collection and forwarding of the specimen described in 2.2.1.2. 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) or legal guardian(s) of the newborn, or in the absence of or inability of the newborn's parent(s) or legal guardian(s), the person in charge of the premises where the birth occurred shall be responsible.

2.3. Care Coordination

The named submitter of an initial newborn screening specimen shall forward any newborn 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 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.

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 Acidemia

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 Acidemia

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 Acidemia

2.4.30 Beta-Ketothiolase Deficiency

2.4.31 Carnitine Uptake Defect

2.4.32 Arginase Deficiency

2.4.33 Malonic Acidemia

2.4.34 Carnitine Palmitoyltransferase Deficiency 1a

2.4.35 Severe Combined Immunodeficiency

2.4.36 Spinal Muscular Atrophy due to homozygous deletion of exon 7 in Survival Motor Neuron 1 gene

2.4.37 Glycogen Storage Disease Type II (Pompe Disease)

2.4.38 Mucopolysacharidosis Type 1 (MPS1)

2.4.39 X-Linked Adrenoleukodystrophy (X-ALD)

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

The parent(s) or other legal guardian(s) of the newborn shall be advised that a second newborn screening test is required for conditions as specified in Rule 3.2.2.2 and 3.3.

3.2.1.1 Births in birthing facilities: it shall be the responsibility of the 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.

3.2.1.2 Births outside birthing facilities: it shall be the responsibility of the physician, registered midwife, or other health professional attending a birth outside a 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 performed.

3.2.2 Collection

3.2.2.1 The attending health care provider shall collect or require the specimen be collected from all newborns at a newborn well child appointment between 8 and 14 days after birth.

The specimen shall consist of capillary blood collected by heel puncture or alternate method authorized by the Laboratory, placed directly 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 demographic slip, all information requested by the Laboratory.

The specimens shall be air dried horizontally for three to four hours. 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.

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 3.3.1, 3.3.3, 3.3.4, 3.3.5 and 3.3.6 unless: an unsatisfactory specimen was submitted for an initial newborn screening specimen; a screen positive result was obtained on an initial newborn screening specimen from the same newborn; there is no record of a satisfactory initial newborn screening specimen submission, or; for 3.3.1 only, the initial newborn screening specimen from the same newborn was collected before 24 hours of life.

3.3 List of Conditions for Second Newborn Screening

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

3.3.5 Cystic Fibrosis

3.3.6 Biotinidase Deficiency

3.3.7 Congenital Adrenal Hyperplasia

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 attempt 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.

SECTION 5: QUALITY CONTROL AND EDUCATION (PROMULGATED BY THE EXECUTIVE DIRECTOR)

5.1 The Laboratory shall have available for review a written quality assurance plan covering testing and reporting initial and second newborn screening specimens.

5.1.1 The written quality assurance plan will include monitoring the positive predictive value of testing.

5.1.1.1 The positive predictive value of testing shall be communicated by the Department to improve the quality of initial and second newborn screening.

5.1.2 The written quality assurance plan will include collecting data on newborns with a screen negative result who are later determined to have a diagnosed condition which was identifiable through screening.

5.2 The Laboratory shall make available educational materials and training concerning initial and second newborn screening specimen collection to all submitting agencies.

5.2.1 The Laboratory shall collect and analyze quality indicators to monitor and inform the newborn screening process including collection and transit of initial newborn screening specimens and unsatisfactory initial newborn screening specimens. The data will be used to implement quality improvement activities.

5.2.1.1 Quality indicators and performance data shall be communicated by the Department to improve the quality of initial and second newborn screening.

Editor's Notes

History

Newborn Screening Regulation 1.6 eff. 01/30/2008.
Second Newborn Screening Regulation 1.6 eff. 04/01/2008.
Entire rule eff. 04/30/2011.
Newborn Screening Regulations 1.1-1.3 eff. 05/30/2012.
Entire rule eff. 08/30/2012.
Rule 1.6 eff. 06/14/2016.
Entire rule eff. 02/14/2019.
Rules 2.4.36, 3.2.2.2 eff. 01/14/2020.
Rules 3.2.2.2, 3.3 emer. rules eff. 04/15/2020; expired 08/13/2020.
Rules 3.2.2.2, 3.3 eff. 08/14/2020.
Rule 2.4 eff. 05/15/2021.