SUMMARY:
These rules and regulations define the responsibilities of hospital administration and staff, physicians, and other health care providers, midwives, and other “principal birthing attendants,” parents and others, with regard to the screening of newborn infants for metabolic disorders and other selected genetic conditions — certain congenital and genetic disorders which, if left untreated, are likely to cause cognitive disabilities or serious illness or death. These rules and regulations address the designation of a contact person in each hospital and birthing center, timing of newborn blood specimen collection, parental/guardian refusal of tests, newborn bloodspot screening (NBS) tests, conditions to be screened, types of records to be maintained, responsibilities for follow-up tests and reporting when necessary, the storage and use of leftover residual NBS specimens; and identifies the fee charged for newborn bloodspot screening NBS.

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SECTION 1.0
PURPOSE

A. These rules and regulations implement 22 M.R.S.A. §§ 1532, 1533 & 22-A M.R.S.A. § 210 which require the Department of Health and Human Services (Department) to: (1) establish a bloodspot screening program for newborns to detect certain congenital genetic disorders which, if left untreated, can cause cognitive, intellectual and developmental disability, serious illness or death, disabilities or serious illness, and (2) establish a statewide genetics program; and (3) authorize the Department to assess fees upon health care providers to cover the costs of bloodspot screening. This rule requires Unless the infant’s parent(s) objects on religious grounds, the responsible hospital, birthing center, physician, midwife, principal birthing attendant, or other health care provider shall cause to be taken to ensure blood NBS specimens are taken from each infant either born in the State of Maine or moving residing in to Maine within three months of birth, unless the infant’s parent(s) object on religious grounds. (see Section 5.1 for infants not born in Maine). The blood specimens shall be obtained by heel stick and collected on filter paper forms available from the Maine Newborn Bloodspot Screening Program, Maine Department of Health and Human Services, and shall be dried and forwarded to the screening laboratory designated by the Department in the envelopes provided within 1 working day after collection.

SECTION 2.0
DEFINITIONS

A. In addition to the definitions listed in 22 M.R.S. § 2150-F, as used in this rule, unless the context indicates otherwise, the following terms have the following meanings:

1.2.1 "Birthing center" means any non-hospital health facility, institution, or place designed to accommodate mothers giving birth away from home at the culmination of normal, uncomplicated pregnancies.

2.2.2 "Principal birthing attendant" means any adult who acts as the principal attendant during a delivery that occurs at a site other than a hospital or birthing center. This may be a midwife or other adult attendant.

3.2.3 "Child Development Services" (CDS) system means an Intermediate Educational Unit that provides both Early Intervention (birth through two years of age) and Free Appropriate Public Education (FAPE for ages three through five years of age) under the supervision of the Maine Department of Education.

4. Core condition means a disorder included in the Recommended Uniform Screening Panel (RUSP) for which Maine NBS specimens must be tested using established laboratory markers/analytes. See Appendix A.

5. Designated currier means a professional delivery service responsible for transporting newborn blood samples daily Monday through Saturday for a contracted laboratory.

6.2.4 "Designated screening laboratory" means the laboratory with which the state Department contracts to process the screening specimens, analyze NBS specimens and to provide report the NBS screening results to the Maine Newborn Screening Program Department.

7.2.5 "Department" means refers to the Maine Department of Health and Human Services (previously known as Department of Human Services), including the Maine Center for...
Disease Control & Prevention (Maine CDC previously known as the Bureau of Health).

8.2.6 Health care provider means a physician, advanced practice nurse, midwife or other licensed professional acting as primary health care provider for the infant.

9. Residual Filter paper means the portion of NBS specimen that remains after screening is complete.

10. Secondary condition means a disorder that can be detected in the differential diagnosis of a core condition. See Appendix A.

11. Time-critical condition means a disorder that may manifest with acute symptoms in the first days of life and require immediate treatment to reduce the risk of morbidity and mortality. See Appendix B.

SECTION 3.0
RESPONSIBILITY FOR SPECIMEN COLLECTION FROM INFANTS BORN IN HOSPITALS OR BIRTHING CENTERS IN MAINE.

A.3.1 The administrator of the hospital or birthing center shall be responsible for ensuring that a blood specimen is collected from each newborn infant prior to his/her discharge from the facility in accordance with Section 7 of this rule for timing of the specimen collection.

B. Parental refusal of the screening tests must be documented and reported in accordance with Section 9 of this rule.

C.3.2 Each administrator of a hospital or birthing center involved in testing under these regulations shall appoint a contact person at the facility who is responsible for coordinating the facility’s screening activities, and provide to the Maine Newborn Screening Program, Division of Family Health Maine Department of Health and Human Services, the name of the contact person at the facility, who shall be responsible for coordinating the facility’s screening activities.

D.3.3 The person who actually draws the blood NBS specimen by performing a heel stick shall fully and clearly complete the filter paper form, and record in the infant’s chart the fact that the blood NBS specimen was collected, including date and time when collected.

E.3.4 No infant shall be discharged until his/her chart is checked to ensure that an NBS blood specimen for newborn screening has been collected. The facility employee who assembles the discharge papers before the infant leaves the facility shall check that an NBS blood specimen has been collected and that this fact has been recorded in the infant’s medical record. The fact that the infant has had a specimen collected shall be included in discharge instructions that are given to the parent(s).

F.3.5 The Maine Newborn Screening Program Department will send to the hospital contact person (see Section 3.2 above) test results for infants whose blood NBS specimens are received for testing. The contact person shall compare these results to the hospital’s list of infants discharged to assure that each infant was tested before discharge, and that each blood specimen was received for testing. If any infant is identified as having been discharged without testing, or without a blood specimen having been received for testing, the contact person shall notify the infant’s physician or other primary health care provider (within 24 hours) and the
Maine Newborn Screening Program Department (within five working days) of discovering that fact. The health care provider shall then take appropriate steps to have the infant tested within five working days of the discovery.

**G.3.6** If an infant is transferred to a second facility during the first 48 hours of life, the blood specimen shall be taken at the second facility. The first facility clearly indicate in the transfer papers accompanying that the infant requires an initial NBS test (see also Section 7.0) and ensure that the Department is notified in writing of the transfer within five working days of the transfer, using the transfer form provided by the Department.

**H.3.7** The administrator of the hospital or birthing center shall ensure that each NBS specimen is forwarded via designated courier to the designated screening laboratory within 1 working day after collection.

**I.3.8** All screening results will be returned by the Maine Newborn Screening Program Department to the hospital contact person (Section 3.2 above), by providing individual result reports. The screening results will be recorded in the individual infant’s medical record.

**J.3.9** The administrator of the hospital or birthing center shall ensure that at least 10 percent of infants’ medical records are reviewed within eight weeks after discharge, to assure that screening information, including result, has been recorded.

**K.3.10** The administrator of the hospital or birthing center shall ensure that all employees are informed of their responsibilities with respect to this rule.

**SECTION 4.0** RESPONSIBILITY FOR NEWBORN BLOODSPOT SPECIMEN COLLECTION FROM INFANTS BORN IN MAINE BUT NOT IN A HOSPITAL OR BIRTHING CENTER.

**A.4.1** If an infant is delivered outside a hospital or birthing center by a midwife or other principal birthing attendant who is authorized to draw blood, that person shall collect an NBS specimen by a heel stick at the appropriate time (in accordance with Section 7.0), complete the filter paper form, and forward it to the designated screening laboratory within 24 hours after collection.

**B.4.2** The midwife or principal birthing attendant shall record in the infant’s record the fact that the specimen was collected, including the date and time of collection, and that the filter paper form was completed and forwarded to the designated screening laboratory.

**C.4.3** Parental refusal of the screening tests must be documented and reported in accordance with Section 9 of this rule.

**D.** If the midwife or principal birthing attendant is not authorized to draw blood, he or she shall:

1.a. Inform the parent(s) about the screening tests and the relevant State law governing them;

2.b. Direct the parent(s) to see an individual authorized to draw blood and have the infant tested by the 3rd day within 24-48 hours of life;

3.e. Contact the parent(s) by the 5th day of life to verify that the infant has been tested; and
4.d. Keep a written record of each of the actions required under the governing statute and this rule.

SECTION 5.0
RESPONSIBILITY FOR NEWBORN BLOODSPOT SPECIMEN COLLECTION
FROM INFANTS NOT BORN IN MAINE

A.5.1 If an infant is not born in the State of Maine but is, or subsequently becomes, a resident of Maine, the first primary health care provider in Maine who examines the infant in the first three months of life should verify whether the infant has been screened, and if no NBS testing has been done, the provider shall submit a heel stick specimen for testing. The health care provider may rely upon the information in the infant’s medical record to determine whether screening has been done.

SECTION 6.0
RESPONSIBILITY OF THOSE PROVIDING PEDIATRIC SERVICES

A.6.1 The primary health care provider licensed to practice in Maine who examines an infant for the first time in the first three months of life and who is subject to this rule should determine whether the child has been screened for causes of cognitive disabilities and selected genetic conditions by checking the infant’s medical records, asking the parent(s) or, if necessary, contacting the Maine Newborn Screening Program. If the health care provider determines that no screening has been performed, the provider shall, within five working days of the initial examination, screen the infant by collecting a blood specimen as outlined in Section 1.0.

B.6.2 Any physician or other health care provider subject to these rules who has identified a case of a child presenting with a genetic condition or metabolic disorder listed in the Department’s Newborn Screening Program shall notify the Newborn Screening Program of such condition within five working days of the identification.

SECTION 7.0
TIMING OF BLOOD SPECIMEN COLLECTION

A.7.1 For term infants, the specimen shall be taken by the 3rd day between 24-48 hours of life or, if the infant’s stay in the hospital or birthing center is less than 3 days, as close to discharge as possible.

B.7.2 For infants who are discharged within 24 hours of birth, a first blood specimen shall be taken as close to discharge from the hospital or birthing center as possible, and a second specimen shall be taken as close to the 3rd day as possible and not later than the 7th day. The administrator of the hospital or birthing center shall ensure the following:

1.a. The infant’s parents are notified of what they need to do to complete the second test;

2.b. The infant’s primary health care provider is notified of the early discharge and of need for the second test; and
3.3. Such notifications are made a part of the infant’s medical records.

C.2.3. For preterm, sick or other infants in intensive care, NBS specimens shall be taken on the day of discharge from the hospital or birthing center or between 24-48 hours of life. If the infant’s stay at the facility is prolonged, beyond 3 days, on the 3rd day of life, regardless of feeding status, with a second NBS specimen must be taken at two weeks, one month and monthly thereafter, or at discharge from intensive care, whichever is earlier, unless otherwise medically indicated.

1. The attending physician providing intensive care to the infant whose stay at the facility extends beyond one month may determine the appropriate frequency of NBS specimen collection and must document this determination in the infant’s medical record and any alternative schedule for collection.

D.2.4. For infants receiving blood transfusions, the NBS specimen shall be taken, if possible, before any anticipated transfusion, regardless of infant’s age. A second NBS specimen shall be taken 3 to 7 days post-transfusion.

SECTION 8.0
SCREENING TEST PERFORMED

A.8.1. The Department will consider changes in conditions to be screened as requested by the Maine Center for Disease Control & Prevention program advisory committee, the medical community or the public. The Department reviews the recommendations from the Advisory Committee on Heritable Disorders in Newborns and Children and the Recommended Uniform Screening Panel (RUSP), and data from medical experts and other newborn screening programs, when considering a new condition. Rulemaking will be conducted in accordance with 5 M.R.S. §§ 8001-11008.

B.8.2. The Department shall determine conditions to be screened, considering whether:

1. The condition has significant mortality and morbidity when not diagnosed before symptoms appear.
2. The condition may not be identified early clinically.
3. The prevalence of the condition in the population is significant.
4. Pre-symptomatic treatment affects outcome.
5. A simple, inexpensive and effective screening method is available; and
6. Resources for treatment and counseling are available.
7. The costs of screening, diagnosis and treatment can be justified by increases in well-being and quality of life for affected individuals and their families.

C.8.3. As of the effective date of these regulations, all newborn blood specimens are must be tested for laboratory markers for the following disorders: conditions specified in this rule. See Appendix A for a list of specified core conditions and secondary conditions.
Biotinidase Deficiency  
Cystic Fibrosis  
Galactosemia  
**Endocrine Disorders:**  
Congenital Adrenal Hyperplasia (CAH)  
Congenital Hypothyroidism  
**Hemoglobinopathies**  
**Amino Acid Disorders:**  
PKU (Phenylketonuria)  
MSUD (Maple Syrup Urine Disease)  
HCU (Homocystinuria)  
Tyr I (Tyrosinemia I)  
Tyr II (Tyrosinemia II)  
**Urea Cycle Disorders:**  
ASS (Citrullinemia)  
ASL (Argininosuccinic Aciduria)  
Argininemia  
HHH Syndrome  
Hyperammonemia  
Hyperornithinemia  
Homocitrullinemia  
**Fatty Acid Oxidation Disorders:**  
MCAD (Medium-Chain Acyl Co-A Dehydrogenase Deficiency)  
LCAD (Long-chain acyl-CoA dehydrogenase deficiency)  
LCHAD (Long-chain hydroxy-CoA dehydrogenase deficiency)  
VLCAD (Very long-chain acyl-CoA dehydrogenase deficiency)  
SCAD (Short chain acyl-CoA dehydrogenase deficiency)  
CPT II (Carnitine Palmitoyl Transferase deficiency Type II (CPT Deficiency))  
GA II (Glutaric Acidemia II)  
CUD (Carnitine Uptake Deficiency)  
TFP (Tri-functional Protein Deficiency)  
**Organic Acid Disorders:**  
GA I (Glutaric Acidemia I)  
IVA (Isovaleric Acidemia)  
MMA (Methylmalonic Aciduria)  
PPA (Propionic Acidemia)  
HMG (HMG CoA Lyase Deficiency)  
MCC (B-Methyl Crotonyl Carboxylase)  
B-KT (B-Ketothiolase Deficiency)  
MCD (Multiple Carnitine Deficiency)  

**SECTION 9.0**  
PARENTAL REFUSAL OF THE SCREENING TESTS

A.9.1 In the instance of parental refusal of the screening tests on religious grounds, the parental refusal shall must be stated in writing and made a part of the infant’s medical record.

B.9.2 The administrator of hospitals and or birthing centers designate, midwife, and principal birthing attendants shall ensure that the Maine Newborn Screening Program, Maine Department of Health and Human Services is notified in writing of the parental refusal within five days of the infant’s birth, using the refusal form provided by the Department.
SECTION 10.0  
FOLLOW-UP TESTS

A. 10.1 The Maine Newborn Screening Program Department shall forward report out-of-range NBS results and follow-up recommendations any follow-up test requests to the infant’s designated primary health-care provider within 2 working days of being notified, by the designated screening laboratory, of the need for follow-up testing, based on the level of urgency and in accordance with this rule. A filter paper form shall be supplied with the follow-up request, unless otherwise indicated.

1. Mildly out-of-range results must be reported within one business day.

2. Moderate and urgent results must be reported on the day the report is received from the laboratory.

   a. The Department’s contract laboratory will report time-critical out-of-range results on weekends and holidays and when the Department is closed.

3. If there is no designated primary health care provider identified, the Department must report to the infant’s parents directly.

B. 10.2 The infant’s designated primary health care provider shall submit a follow-up test NBS specimen, within the timeframe recommended by the Department. The provider must obtain other laboratory tests specified by the Department using the supplied filter paper form, within 5 working days of the date of the request, or consult with pediatric specialty consultant within the timeframe specified by the Department as otherwise indicated in the request.

C. 10.3 If the infant’s designated health care provider cannot submit a follow-up test specimen within 5 working days the specified timeframe, the provider shall notify the Maine Newborn Screening Program Department of this fact and the reason for it.

D. 10.4 If the infant’s designated health care provider processes a requested repeat specimen through a local laboratory, the provider will notify the Department of the results.

E. 10.5 Test results for repeat or follow-up screening tests will be reported directly to the appropriate health care provider by providing individual result reports.

F. 10.6 For the purpose of coordinating efforts to detect, prevent, and treat genetic conditions and metabolic disorders, the Department may share individually identifiable health information related to the potential or actual presence of the genetic conditions and metabolic disorders, that are listed in this rule, with other public health programs and agencies whose mission is to detect, prevent and treat these disorders, including Child Development Services (CDS).
SECTION 11.0
ADVISORY COMMITTEE

A. The Department shall appoint a program advisory committee, the Joint Advisory Committee for the Identification and Management of Children with Conditions Detected through the Maine Newborn Bloodspot Screening Program (JAC), to advise the program on issues related to the screening and follow-up of newborns.

SECTION 12.0
FILTER PAPER STORAGE AND USE

A. The primary use of filter paper specimens is for the processing of newborn screening tests as allowed by the rule. Residual specimens are used to support essential program functions such as program evaluation, quality assurance, result verification, test refinement, and quality improvement initiatives. Specifically, residual NBS specimens are used to document that specimens were properly collected, transported, received and analyzed for the benefit of the newborn.

B. After testing is completed, the contracted laboratory will store leftover residual filter paper specimens will be stored indefinitely. This policy shall be reviewed by the Advisory Committee, every five years and recommendations made to the Department. The contracted laboratory storage must be in accordance with Clinical and Laboratory Standards Institute Guidelines (CLSI) (https://clsi.org/). Storage conditions shall be appropriate, secure and stable, and storage must allow specimens to be retrieved, if necessary.

C. Leftover residual filter paper specimens may be used for further testing as indicated or requested by the submitting or attending health-care provider if these tests are available through the contracted laboratory or through other laboratories. The healthcare provider must obtain verbal consent from the parent or legal guardian, and complete and sign the Request for Retrieval of Newborn Filter Paper Specimens for Additional Testing Form provided by the Department at the request of the healthcare provider.

D. The information collected in this program is maintained by the Department. Information is used to identify infants at risk of developing cognitive-intellectual and developmental disorders or serious illness in order to develop programs to prevent and detect such disorders.

E. Unless the person or his/her legal authorized representative specifically prohibits such use in writing, the blood specimen and information obtained during the testing process becomes the property of the State Department and may be used for program evaluation or research by the Department or Department-approved scientific researchers to improve the health of mothers and children. Such studies are published without identifying the person or persons from whom these results were obtained.

F. The Department, with input from the JAC, may release de-identified NBS samples to external agencies for research projects if the research project has received approval through the Department’s process for research requests.

1. Prior to release of NBS, samples must be de-identified and assigned a unique numeric identifier by the Department to prevent the sample from being linked to
the original specimen, unless the Department has written record of parental consent to release identifiable information; and

2. Any letters of support provided by the Department to an external agency will stipulate that released samples are limited in use for the specifically approved study.

3. Any remaining residual NBS samples in the custody of researchers must be destroyed by the research program upon completion of the study.

12.7 Filter paper specimens may be released for research or testing with identifiers intact with specific written request or consent of a parent/guardian; for anonymous research without consent as approved by the Department with input from the program advisory committee; or for program evaluation or planning without consent.

SECTION 13.
FILTER PAPER DESTRUCTION

A. Residual filter paper specimens may be destroyed at the request of an individual and parent or guardian of a child. Written requests for specimen destruction must be submitted in writing using the Request for Destruction of Newborn Filter Paper Specimen Form provided by the Department.

B. Residual filter paper specimens may be destroyed at the request of the Department if the filter paper specimens have been affected by storage conditions inconsistent with the current guidelines issued by the CLSI that develops standards to assist laboratories to fulfill the responsibilities of newborn bloodspot screening with efficiency and effectiveness. Refer to http://clsi.org/standards/.

SECTION 14.12.0
FEES

A.13.1 The Department charges a fee for the newborn bloodspot screening tests. As of the effective date of these regulations, the fee is $110 per infant tested. Hospitals and health care providers are charged this fee by the purchase of the filter paper form used for blood collection.

B.13.2 Hospitals and health care providers will receive a credit on future filter paper orders for repeat specimens that have been submitted. To receive the credit, hospitals and providers must submit to the Maine Newborn Screening Program a list of infants for whom repeat specimens were obtained.

SECTION 15.14.0
PENALTIES

A. Failure to comply with these regulations may result in the imposition of such civil and criminal penalties as are specified under 22 M.R.S.A., §Section 47.
STATUTORY AUTHORITY: 22 M.R.S.A. §§ 1532 and 1533

EFFECTIVE DATE:
   April 15, 1984

AMENDED:
   November 24, 1986
   August 1, 1995

EFFECTIVE DATE (ELECTRONIC CONVERSION):
   May 5, 1996

AMENDED:
   October 1, 1998
   May 22, 2006 – filing 2006-205
   December 1, 2009 – filing 2009-612
### APPENDIX A.

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<th>Core Conditions</th>
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<td>2-Methylbutyrylglycinuria (2MBG)</td>
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<td>2-Methyl-3-Hydroxybutyryl Aciduria (2M3HBA)</td>
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<td>T-cell Related Lymphocyte Deficiencies</td>
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<td>Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)</td>
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