

**MAINE DEPARTMENT OF HEALTH AND HUMAN SERVICES
MAINE CENTER FOR DISEASE CONTROL & PREVENTION
DIVISION OF FAMILY HEALTH
NEWBORN SCREENING PROGRAM**

CHAPTER 283

**RULES AND REGULATIONS RELATING TO TESTING NEWBORN INFANTS
FOR DETECTION OF CAUSES OF MENTAL RETARDATION AND
SELECTED GENETIC CONDITIONS**

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 inborn errors of metabolism and other selected genetic conditions. These rules and regulations address the designation of a contact person in each hospital, timing of newborn blood specimen collection, parental refusal of tests, conditions to be screened, types of records to be maintained, responsibilities for follow-up tests and reporting when necessary, and the storage and use of leftover specimens.

1.0 PURPOSE

These rules and regulations implement section 1532 of Title 22 of the Maine Revised Statutes Annotated, governing the testing of newborn infants for the detection of causes of mental retardation and section 1533 of Title 22 of the Maine Revised Statutes Annotated, establishing a statewide genetics program. Unless the infant’s parent(s) objects on religious grounds, the responsible hospital, birthing center, physician, midwife, principal birthing attendant, or health care provider shall cause to be taken blood specimens from each infant either born in the State of Maine or moving to Maine within three months of birth (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 Health and Environmental Testing Laboratory (HETL), 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 with the forms within 1 working day after collection.

2.0 DEFINITIONS

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 “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.
- 2.3 “Designated screening laboratory” means the laboratory with which the state contracts to process the screening specimens and to provide the screening results to the Maine Newborn Screening Program.
- 2.4 “Department” 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 (previously known as the Bureau of Health).
- 2.5 “Health care Provider” means a physician, advanced practice nurse or other licensed professional acting as primary health care provider for the infant.

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

- 3.1 The administrator of the hospital/birthing center shall be responsible for assuring that a blood specimen is collected from each newborn infant prior to his/her discharge from the facility (see Section 7 for timing of the specimen collection).
- 3.2 Each administrator of a hospital or birthing center involved in such testing shall appoint, and provide to the Maine Newborn Screening Program, Division of Family Health, Maine Department of Health and Human Services, the name of a contact person at the facility, who shall be responsible for coordinating the facility’s screening activities.
- 3.3 The person who actually draws the blood 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 specimen was collected, including date and time when collected.
- 3.4 No infant shall be discharged until his/her chart is checked to assure that a 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 a 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 any discharge instructions that are given to the parent(s).
- 3.5 The Maine Newborn Screening Program will send, to the hospital contact person (see Section 3.2 above) test results for infants whose blood

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 health care provider (within 24 hours) and the Maine Newborn Screening Program (within 5 working days) of discovering that fact. The health care provider shall then take appropriate steps to have the infant tested within 5 working days.

- 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 shall clearly indicate in the papers accompanying the infant that the child needs to be screened (see also Section 7.0) and notify the Maine Newborn Screening Program of the transfer within 5 working days.
- 3.7** The administrator of the hospital or birthing center shall ensure that each blood specimen is forwarded to the designated screening laboratory within 1 working day after collection.
- 3.8** All screening results will be returned by the Maine Newborn Screening Program to the hospital contact person (Section 3.2 above), by providing individual result reports. The screening results will be recorded in the individual infants medical record.
- 3.9** The administrator of the hospital or birthing center shall ensure that at least 10% of infants' medical records are reviewed within 8 weeks after discharge, to assure that screening information, including result, has been recorded.
- 3.10** The administrator of the hospital or birthing center shall ensure that all employees are informed of their responsibilities with respect to these regulations.

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

- 4.1** If an infant is delivered outside a hospital or birthing center, midwife or the principal birthing attendant who is authorized to draw blood, shall ensure that a blood specimen is collected by performing a heel stick at the appropriate time (Section 7.0), dried and that the filter paper form is fully completed and forwarded to the designated screening laboratory within 1 working day after collection of the specimen.

- 4.2** This midwife or principal birthing attendant shall record in the infant's record the fact that the blood specimen was collected, including date and time of collection, and that the filter paper form was completed and forwarded to the designated screening laboratory.
- 4.3** If the midwife or principal birthing attendant is not authorized to draw blood, he or she shall:
- a. inform the parent(s) about the screening tests and the State law governing them;
 - b. direct the parent(s) to see an individual authorized to draw blood and have the infant tested by the 3rd day of life;
 - c. contact the parent(s) by the 5th day of life to verify that the infant has been tested; and
 - d. keep a written record of each of the actions required under this rule.

5.0 RESPONSIBILITY FOR SPECIMEN COLLECTION FROM INFANTS NOT BORN IN MAINE

- 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 3 months of life should verify whether the infant has been screened, and if not, shall perform the test. The health care provider may rely upon the information in the infant's medical record to determine whether such screening has been done.

6.0 RESPONSIBILITY OF THOSE PROVIDING PEDIATRIC SERVICES

- 6.1** The primary health care provider in Maine who examines an infant for the first time in the first three months of life should determine whether the child has been screened for causes of mental retardation 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, he/she shall, within 5 working days, screen the infant by collecting a blood specimen as outlined in Section 1.0.
- 6.2** Any physician or other health care provider subject to these rules who has identified a **(potential)** 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 business days of the identification.

7.0 TIMING OF BLOOD SPECIMEN COLLECTION

- 7.1** For term infants, the specimen shall be taken by the 3rd day 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.
- 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 assure:
- a. that the infant's parents are notified of what they need to do to complete the second test;
 - b. that the infants' primary health care provider is notified of the early discharge and of need for the second test; and
 - c. that such notifications are made a part of the infants' medical records.
- 7.3** For preterm, sick or other infants in intensive care, specimens shall be taken on the day of discharge from the hospital or birthing center or, if the stay at the facility is prolonged beyond 3 days, on the 3rd day of life, regardless of feeding status with a second specimen taken at 2 weeks or at discharge from intensive care, whichever is earlier, unless otherwise indicated.
- 7.4** For infants receiving blood transfusions, obtain specimen, if possible, before any anticipated transfusion, regardless of infant's age. Obtain a second specimen 3 to 7 days post-transfusion.

8.0 SCREENING TEST PERFORMED

- 8.1** The Department will consider changes in conditions to be screened as requested by the Maine Center for Disease Control & Prevention, the medical community or the public. The Department shall consult with medical providers and the program advisory committee in making these decisions.
- 8.2** The Department shall determine conditions to be screened considering:
- The condition has significant mortality and morbidity when not diagnosed before symptoms appear.
 - The condition may not be identified early clinically.
 - The prevalence of the condition in the population is significant.
 - Presymptomatic treatment affects outcome.
 - A simple, inexpensive and effective screening method is available.
 - Resources for treatment and counseling are available.
 - 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.

- 8.3** As of the effective date of these regulations, all newborn blood specimens are tested for the following disorders:

Biotinidase Deficiency

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)

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)

9.0 PARENTAL REFUSAL OF THE SCREENING TESTS

- 9.1** In the instance of parental refusal of the screening tests on religious grounds, the parental refusal shall be stated in writing and made a part of the infant's medical record.
- 9.2** The administrator of hospitals and birthing centers, 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 14 days of the infant's birth.

10.0 FOLLOW-UP TESTS

- 10.1** The Maine Newborn Screening Program shall forward any follow-up test requests to the appropriate health care provider within 2 working days of being notified, by the designated screening laboratory, of the need for follow-up testing. A filter paper form shall be supplied with the follow-up request, unless otherwise indicated.
- 10.2** The health care provider shall submit a follow-up test specimen, using the supplied filter paper form, within 10 working days of the date of the request, or as otherwise indicated in the request.
- 10.3** If the health care provider cannot submit a follow-up test specimen within 10 working days, he/she shall notify the Maine Newborn Screening Program of this fact and the reason for it.
- 10.4** If the health care provider processes a requested repeat specimen through a local laboratory, he/she will notify the Maine Newborn Screening Program of the results.
- 10.5** Test results for repeat or follow-up screening tests will be reported directly to the appropriate health care provider by providing result reports.
- 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 presence of genetic conditions and metabolic disorders, listed in the Department's Newborn Screening Program with other public health programs and agencies whose mission is to detect, prevent and treat these disorders.

11.0 ADVISORY COMMITTEE

- 11.1** The Department shall appoint an advisory committee to advise the program on issues related to the screening and follow-up of newborns.

12.0 FILTER PAPER STORAGE AND USE

- 12.1** The primary use of filter paper specimens is for the processing of newborn screening tests as allowed by these rules.
- 12.2** After testing is completed, leftover 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. Storage conditions shall be appropriate, secure and stable and allow specimens to be retrieved if necessary.

- 12.3 Leftover filter paper specimens may be used for further testing as indicated/requested by the submitting or attending health care provider if these tests are available through contracted laboratory or through other laboratories with consent of the family.
- 12.5 The information collected in this program is maintained by the Department. Information is used to identify infants at risk of birth defects in order to develop programs to prevent and detect such defects.
- 12.6 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 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.
- 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.

13.0 PENALTIES

- 13.1 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.

BASIS STATEMENT: These rules were adopted to define responsibilities to assure that all infants born in Maine are screened for causes of mental retardation and selected genetic conditions (unless the infant's parent(s) object on religious grounds) in time to allow for treatment to prevent retardation and other health problems.

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