

**STATE OF MAINE**  
**NEWBORN BLOODSPOT SCREENING RULE**

**10-144 CODE OF MAINE RULES**  
**CHAPTER 283**



Department of Health and Human Services  
Maine Center for Disease Control and Prevention  
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Augusta, Maine 04333-0011

**ROUTINE TECHNICAL RULE**  
**Effective June 1, 2021**

**PROVISIONALLY ADOPTED MAJOR SUBSTANTIVE RULE**  
**May 25, 2021**

**10-144: MAINE DEPARTMENT OF HEALTH AND HUMAN SERVICES  
MAINE CENTER FOR DISEASE CONTROL AND PREVENTION**

**Chapter 283: NEWBORN BLOODSPOT SCREENING RULE**

**NOTICE**

This rule is a routine technical rule, EXCEPT for Section 14 (Fees), which is a major substantive rule, pursuant to 5 M.R.S. §8071(3)(B).

**SUMMARY**

This rule defines the responsibilities of hospital administration and staff, physicians, healthcare providers, midwives, principal birthing attendants, parents and others; with regard to the screening of newborn infants for certain congenital and genetic disorders which, if left untreated, can lead to intellectual and developmental disability, serious illness or death. This rule addresses the designation of a contact person in each hospital and birthing center, timing of newborn blood specimen collection, parental/guardian refusal of newborn bloodspot screening (NBS) tests, conditions to be screened, types of records to be maintained, responsibilities for follow-up tests and reporting when necessary, storage and use of residual filter paper specimens; and identifies the fee charged for NBS.

**TABLE OF CONTENTS**

<b>SECTION</b>	<b>PAGE</b>
<b>SECTION 1</b> PURPOSE.....	<b>1</b>
<b>SECTION 2</b> DEFINITIONS.....	<b>1</b>
<b>SECTION 3</b> RESPONSIBILITY FOR SPECIMEN COLLECTION FROM INFANTS BORN IN HOSPITALS OR BIRTHING CENTERS IN MAINE.....	<b>2</b>
<b>SECTION 4</b> RESPONSIBILITY FOR NEWBORN BLOODSPOT SPECIMEN COLLECTION FROM INFANTS BORN IN MAINE BUT NOT IN A HOSPITAL OR BIRTHING CENTER.....	<b>3</b>
<b>SECTION 5</b> RESPONSIBILITY FOR NEWBORN BLOODSPOT SPECIMEN COLLECTION FROM INFANTS NOT BORN IN MAINE.....	<b>3</b>
<b>SECTION 6</b> RESPONSIBILITY OF THOSE PROVIDING PEDIATRIC SERVICES.....	<b>3</b>
<b>SECTION 7</b> TIMING OF BLOOD SPECIMEN COLLECTION.....	<b>4</b>
<b>SECTION 8</b> SCREENING TEST PERFORMED.....	<b>4</b>

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<b>SECTION 9</b>	
PARENTAL REFUSAL OF THE SCREENING TESTS.....	<b>5</b>
<b>SECTION 10</b>	
FOLLOW-UP TESTS.....	<b>5</b>
<b>SECTION 11</b>	
ADVISORY COMMITTEE.....	<b>6</b>
<b>SECTION 12</b>	
RESIDUAL FILTER PAPER SPECIMEN STORAGE AND USE.....	<b>6</b>
<b>SECTION 13</b>	
RESIDUAL FILTER PAPER SPECIMEN DESTRUCTION.....	<b>7</b>
<b>SECTION 14</b>	
FEES.....	<b>7</b>
<b>SECTION 15</b>	
PENALTIES.....	<b>8</b>
<b>STATUTORY AUTHORITY.....</b>	<b>8</b>
<b>APPENDIX A.....</b>	<b>9</b>
<b>APPENDIX B.....</b>	<b>10</b>

## SECTION 1 PURPOSE

- A. This rule implements 22 MRS §§ 1532,1533 and 22-A MRS §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 intellectual and developmental disability, serious illness or death, and (2) establish a statewide genetics program; and (3) authorize the Department to assess fees upon healthcare providers to cover the costs of bloodspot screening. This rule requires the responsible hospital, birthing center, physician, midwife, principal birthing attendant, or other healthcare provider to ensure NBS specimens are taken from each infant either born in the State of Maine or residing in Maine within three months of birth, unless the infant's parent(s) object on religious grounds.

## SECTION 2 DEFINITIONS

- A. As used in this rule, unless the context indicates otherwise, the following terms have the following meanings:
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. **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.
  3. **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.
  4. **Designated courier** means a professional delivery service responsible for transporting newborn blood samples daily, Monday through Saturday, for a contracted laboratory.
  5. **Designated screening laboratory** means the laboratory with which the Department contracts to analyze NBS specimens and report the NBS screening results to the Department.
  6. **Department** means the Department of Health and Human Services Maine Center for Disease Control and Prevention.
  7. **Healthcare provider** means a physician, advanced practice nurse, midwife or other licensed professional acting as primary healthcare provider for the infant.
  8. **Principal birthing attendant** means any adult who acts as the primary attendant during a delivery that occurs at a site other than a hospital or birthing center.
  9. **Residual filter paper specimen** 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**  
**RESPONSIBILITY FOR SPECIMEN COLLECTION FROM**  
**INFANTS BORN IN HOSPITALS OR BIRTHING CENTERS IN MAINE**

- A. The administrator of the hospital or birthing center is responsible for ensuring that a blood specimen is collected from each newborn infant prior to his/her discharge from the facility in accordance with this rule.
- B. Each administrator of a hospital or birthing center involved in testing under this rule must designate a contact person at the facility who is responsible for coordinating the facility's screening activities, and provide to the Department the name of the contact person responsible for coordinating the facility's screening activities.
- C. The person who collects the NBS specimen by performing a heel stick must fully and clearly complete the filter paper form, and record in the infant's chart the fact that the NBS specimen was collected, including date and time when collected.
- D. No infant may be discharged until his/her chart is checked to ensure that an NBS specimen has been collected. The facility employee who assembles the discharge papers before the infant leaves the facility must check that an NBS specimen has been collected and recorded in the infant's medical record. The fact that the infant has had a specimen collected must be included in discharge instructions that are given to the parent(s).
- E. The Department will send to the hospital contact person test results for infants whose NBS specimens are received for testing. The contact person must compare these results to the hospital's list of infants discharged to ensure 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 NBS having been received for testing, the contact person must notify the infant's physician or other primary healthcare provider within 24 hours and the Department within five working days of discovering that fact. The healthcare provider must then take appropriate steps to have the infant tested within five working days of the discovery.
- F. If an infant is transferred to a second facility during the first 24 hours of life, the NBS specimen must be taken at the second facility. The first facility must clearly indicate in the transfer papers accompanying that the infant requires an initial NBS test. The transferring hospital must 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.
- G. The administrator of the hospital or birthing center must ensure that each NBS specimen is shipped via designated courier to the designated screening laboratory within 24 hours after collection.
- H. All screening results will be returned by the Department to the hospital contact person (Section 3(C) above), by providing individual result reports. The screening results will be recorded in the individual infant's medical record.

- I. The administrator of the hospital or birthing center must 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.
- J. The administrator of the hospital or birthing center must ensure that all employees are informed of their responsibilities with respect to this rule.

**SECTION 4**  
**RESPONSIBILITY FOR NEWBORN BLOODSPOT SPECIMEN COLLECTION**  
**FROM INFANTS BORN IN MAINE BUT NOT IN A HOSPITAL OR BIRTHING CENTER**

- A. If an infant is delivered outside a hospital or birthing center by a midwife or principal birthing attendant who is qualified to draw blood, that person must collect an NBS specimen by a heel stick in accordance with Section 7, complete the filter paper form, and ship the specimen and form to the designated screening laboratory within 24 hours after collection of the specimen.
- B. The midwife or principal birthing attendant must record in the infant's record the fact that the NBS 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. If the midwife or principal birthing attendant is not qualified to draw blood, he or she must:
  - 1. Inform the parent(s) about the screening tests and the relevant State law;
  - 2. Direct the parent(s) to see an individual qualified to draw blood and have the infant tested within 24-48 hours of life;
  - 3. Contact the parent(s) by the third day of life to verify that the infant has been tested; and
  - 4. Keep a written record of each of the actions required under this rule.

**SECTION 5**  
**RESPONSIBILITY FOR NEWBORN BLOODSPOT SPECIMEN COLLECTION**  
**FROM INFANTS NOT BORN IN MAINE**

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

**SECTION 6**  
**RESPONSIBILITY OF THOSE PROVIDING PEDIATRIC SERVICES**

- A. The primary healthcare 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 must determine whether the child has received an NBS test by checking the infant's medical records, asking the parent(s) or, if necessary, contacting the Department. If the healthcare provider determines that no screening has been performed, the provider must, within five working days of the initial examination, screen the infant by collecting a blood specimen as outlined in this rule.

- B.** Any physician or other healthcare provider subject to this rule who has identified a case of a child presenting with a genetic condition or metabolic disorder listed in this rule must notify the Department of such condition within five working days of the identification.

**SECTION 7  
TIMING OF BLOOD SPECIMEN COLLECTION**

- A.** For term infants, the specimen must be taken between 24-48 hours of life.
- B.** For infants who are discharged within 24 hours of birth, a first blood specimen must be taken as close to discharge from the hospital or birthing center as possible, and a second specimen must be taken between 24-48 hours of life. The administrator of the hospital or birthing center must ensure the following:
1. The infant's parents are notified of what they need to do to complete the second test;
  2. The infant's primary healthcare provider is notified of the early discharge and of need for the second test; and
  3. Such notifications are made a part of the infant's medical records.
- C.** For preterm, sick or other infants in intensive care, NBS specimens must be taken between 24-48 hours of life. If the infant's stay at the facility is prolonged, 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.** For infants receiving blood transfusions, the NBS specimen must be taken, if possible, before any anticipated transfusion, regardless of infant's age. A second NBS specimen must be taken 48 hours post-transfusion.

**SECTION 8  
SCREENING TEST PERFORMED**

- A.** The Department will consider changes in conditions to be screened as requested by the Joint 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 to add conditions will be conducted in accordance with 5 MRS §§ 8001-11008.
- B.** The Department must 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. As of the effective date of this rule, all newborn blood specimens must be tested for laboratory markers for the conditions specified in this rule. See Appendix A for a list of specified core conditions and secondary conditions.

### **SECTION 9 PARENTAL REFUSAL OF THE SCREENING TESTS**

- A. In the instance of parental refusal of the screening tests on religious grounds, the parental refusal must be stated in writing and made a part of the infant's medical record.
- B. The hospital or birthing center designee, midwife, and principal birthing attendants must ensure that the Department 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 FOLLOW-UP TESTS**

- A. The Department must report out-of-range NBS results and follow-up recommendations to the infant's designated primary healthcare provider based on the level of urgency and in accordance with this rule.
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 healthcare provider identified, the Department must report to the infant's parent(s) directly.
- B. The infant's designated primary healthcare provider must submit a follow-up NBS specimen within the timeframe recommended by the Department. The provider must obtain other laboratory tests specified by the Department and/or consult with pediatric specialty consultant within the timeframe specified by the Department.
- C. If the infant's designated healthcare provider cannot submit a follow-up test specimen within the specified timeframe, the provider must notify the Department of this fact and the reason for it.



- D. If the infant's designated healthcare provider processes a requested repeat specimen through a local laboratory, the provider will notify the Department of the results.
- E. Test results for repeat or follow-up screening tests will be reported directly to the appropriate healthcare provider by providing individual result reports.
- F. 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 CDS.

### **SECTION 11 JOINT ADVISORY COMMITTEE (JAC)**

- A. The Department shall appoint a program advisory committee, the Joint Advisory Committee for Maine Newborn Screening (JAC), to advise the program on issues related to the screening, including the retention and use of residual filter paper specimens, and follow-up services.
- B. Meetings are generally held twice per year and are open to the public. Department notifications of JAC meeting dates and times may be found at <https://www.maine.gov/dhhs/mecdc/population-health/mch/cshn/bloodspot-screening/index.html>.

### **SECTION 12 RESIDUAL FILTER PAPER SPECIMEN STORAGE AND USE**

- A. The primary use of residual filter paper specimens is for the processing of newborn screening tests as allowed by this rule. Residual filter paper specimens are used to support essential program functions such as program evaluation, quality assurance, result verification, test refinement, and quality improvement initiatives. Specifically, residual filter paper 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 residual filter paper specimens indefinitely. The contracted laboratory storage must be in accordance with Clinical and Laboratory Standards Institute Guidelines (CLSI) ( <https://clsi.org/> ). Storage conditions must be appropriate, secure and stable, and storage must allow specimens to be retrieved, if necessary.
- C. Residual filter paper specimens may be used for further testing as recommended by the healthcare provider if these tests are available through the contracted laboratory or through other laboratories. The healthcare provider must obtain a signed (Department) Authorization to Release Information from the parent/guardian or the individual if they are a legal adult. The healthcare provider must also complete and sign the Request for Retrieval of Residual Filter Paper Specimens for Additional Testing Form provided by the Department at the request of the healthcare provider. The costs associated with additional testing that is not required by this rule is the responsibility of the parent or legal guardian.
- D. The information collected in this program is maintained by the Department. Information is used to identify infants at risk of developing intellectual and developmental disabilities or serious illness and to develop programs to prevent and detect such disorders.

- E.** Information obtained during the testing process becomes the property of the Department and may be used in compliance with confidentiality laws, for program evaluation or research by the Department or Department-approved scientific researchers to improve the health of mothers and children.
- F.** Newborn blood specimens obtained during the testing process become the property of the Department and may be used in compliance with confidentiality laws, for program evaluation or research by the Department or Department-approved scientific researchers to improve the health of mothers and children, unless the person or his/her legal authorized representative:
1. Specifically prohibits such use in writing on a form provided by the Department; or
  2. Requests destruction of the residual filter paper specimen under Section 13.
- G.** The Department, with input from the JAC, may release de-identified residual filter paper specimens or 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 any residual filter paper specimens or samples, the Department must de-identify specimens or samples by assigning a unique numeric identifier to prevent the specimen or sample from being linked to the original specimen or sample, unless the Department has written record of parental consent to release identifiable information; and
  2. Prior to release of any residual filter paper specimens or samples, the external agency receiving the specimens or samples will sign an appropriate Confidentiality and Use Agreement which will specify that the released specimen or sample and any information obtained with, or derived from, it may only be used for the specifically approved study.
  3. Any remaining residual filter specimens or samples in the custody of researchers must be destroyed by the research program upon completion of the study.

**SECTION 13  
RESIDUAL FILTER PAPER SPECIMEN 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 Residual Filter Paper Specimen Form provided by the Department.
- B.** Residual filter paper specimens may be destroyed at the request of the Department if the residual 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 IS A MAJOR SUBSTANTIVE RULE  
SECTION 14  
FEES**

- A.** Hospitals and healthcare providers must use the filter paper approved by the Department for NBS specimen collection.
- B.** The Department charges a fee of \$220 per infant tested, by pricing the filter paper used for each infant at \$220.

- C. Hospitals and healthcare 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 a list of infants for whom repeat specimens were obtained, to the Department.

**SECTION 15  
PENALTIES**

- A. Failure to comply with this rule may result in the imposition of such civil and criminal penalties as are specified under 22 MRS §47.
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**STATUTORY AUTHORITY:**

22 MRS §§ 1532 and 1533; 22-A MRS §210

**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

November 14, 2018 – filing 2018-236

March 11, 2021 – filing 2021- Emergency Routine Technical/Emergency Major Substantive Rule

**PROVISIONALLY ADOPTED MAJOR SUBSTANTIVE FILING:**

May 25, 2021 - filing LR-2021-8

**AMENDED:**

June 1, 2021 – Routine Technical adoption – filing 2021-118

**APPENDIX A**

<b>Core Conditions</b>	<b>Secondary Conditions</b>
<ul style="list-style-type: none"> <li>• 3-Hydroxy-3-Methylglutaric Aciduria (HMG)</li> <li>• 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)</li> <li>• Argininosuccinic Aciduria (ASA)</li> <li>• B-Ketothiolase Deficiency (BKT)</li> <li>• Biotinidase Deficiency (BIOT)</li> <li>• Carnitine uptake Defect/Carnitine Transport Defect (CUD)</li> <li>• Citrullinemia Type I (CIT)</li> <li>• Classic Galactosemia (GALT)</li> <li>• Classic Phenylketonuria (PKU)</li> <li>• Congenital Adrenal Hyperplasia (CAH)</li> <li>• Congenital Hypothyroidism (CH)</li> <li>• Cystic Fibrosis (CF)</li> <li>• Glutaric Acidemia Type I (GAI)</li> <li>• Holocarboxylase Synthase Deficiency (MCD)</li> <li>• Homocystinuria (HCY)</li> <li>• Isovaleric Acidemia (IVA)</li> <li>• Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)</li> <li>• Maple Syrup Urine Disease (MSUD)</li> <li>• Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)</li> <li>• Methylmalonic Acidemia (MUT)</li> <li>• Methylmalonic Acidemia Cobalamin A, B (Cbl A, B)</li> <li>• Mucopolysaccharidosis Type 1 (MPS-1)</li> <li>• Pompe Propionic Acidemia (PROP)</li> <li>• S,C Disease (Hb S/C)</li> <li>• S/ <math>\beta</math>Thalassaemia (Hb S/ <math>\beta</math>Th)</li> <li>• Severe Combined Immunodeficiencies (SCID)</li> <li>• Spinal Muscular Atrophy (SMA)</li> <li>• S,S Disease (Sickle Cell Anemia) (Hb SS)</li> <li>• Trifunctional Protein Deficiency (TFP)</li> <li>• Tyrosinemia Type I (TYR I)</li> <li>• Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)</li> <li>• X-linked Adrenoleukpdystrophy (X-ALD)</li> </ul>	<ul style="list-style-type: none"> <li>• 2-Methylbutyrylglycinuria (2MBG)</li> <li>• 2-Methyl-3-Hydroxybutyric Aciduria (2M3HBA)</li> <li>• 3-Methylglutaconic Aciduria (3MGA)</li> <li>• Argininemia (ARG)</li> <li>• Benign Hyperphenylalaninemia (H-PHE)</li> <li>• Biopterin Defect in Cofactor Biosynthesis (BIOPT (BS))</li> <li>• Biopterin Defect in Cofactor Regeneration (BIOPT (REG))</li> <li>• Carnitine Acylcarnitine Translocase Deficiency (CACT)</li> <li>• Carnitine Palmitoyltransferase Type II Deficiency (CPT II)</li> <li>• Carnitine Palmitoyltransferase Type I Deficiency (CPT 1A)</li> <li>• Citrullinemia, Type II (CIT II)</li> <li>• Galactokinase Deficiency (GALK)</li> <li>• Galactoepimerase Deficiency (GALE)</li> <li>• Glutaric Acidemia Type II (GA2)</li> <li>• Hypermethioninemia (MET)</li> <li>• Isobutyrylglycinuria (IBG)</li> <li>• Medium-chain Ketoacyl-CoA Thiolase Deficiency (MCAT)</li> <li>• Methylmalonic acidemia with homocystinuria (Cbl C,D)</li> <li>• T-cell Related Lymphocyte Deficiencies</li> <li>• Tyrosinemia, Type II (TYR II)</li> <li>• Tyrosinemia, Type III (TYR III)</li> <li>• Various other hemoglobinopathies (Var Hb)</li> </ul>

**APPENDIX B**

<b>Time-critical Conditions</b>
<ul style="list-style-type: none"><li>• 3-Hydroxy-3-Methylglutaric Aciduria (HMG)</li><li>• Argininosuccinic Aciduria (ASA)</li><li>• B-Ketothiolase Deficiency (BKT)</li><li>• Citrullinemia Type I (CIT)</li><li>• Classic Galactosemia (GALT)</li><li>• Congenital Adrenal Hyperplasia (CAH)</li><li>• Glutaric Acidemia Type I (GAI)</li><li>• Holocarboxylase Synthase Deficiency (MCD)</li><li>• Isovaleric Acidemia (IVA)</li><li>• Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)</li><li>• Maple Syrup Urine Disease (MSUD)</li><li>• Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)</li><li>• Methylmalonic Acidemia (MUT)</li><li>• Propionic Acidemia (PROP)</li><li>• Trifunctional Protein Deficiency (TFP)</li><li>• Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)</li></ul>