

## **Newborn Screening Program Rule**

### **1.0 Authority**

This rule is adopted pursuant to 3 V.S.A. § 3003; 18 V.S.A. §§ 102, 115 and 5087.

### **2.0 Purpose and Scope**

The purpose of this rule is to provide standards for screening for certain diseases in newborn children where early identification and treatment may prevent severe disability and/or death by assuring timely initiation of treatment services. These screenings are part of the early case-finding program for chronic diseases. This rule applies to all health care providers for newborn infants.

### **3.0 Definitions**

- 3.1 “Department” means the Vermont Department of Health.
- 3.2 “Dried bloodspot” means the blood specimen drawn for the purpose of screening newborns for certain serious disorders not readily apparent at birth and which require early diagnosis and treatment.
- 3.3 “Newborn Screening Program” means the Department of Health’s program to assure that infants born in the state are tested for certain diseases and conditions for which early identification and treatment will prevent severe disability and/or death, and, for those affected, to assure timely initiation of treatment services.
- 3.4 “Newborn screening test” means the screening for certain rare and serious diseases and conditions which may not be apparent at birth. Newborn screening tests are conducted through a laboratory analysis of dried bloodspots obtained from newborn infants and point-of-care testing.
- 3.5 “Point-of-care testing” means physiologic tests that are administered to newborns and interpreted at the bedside, the site of direct delivery of health care, such as hearing screening to detect hearing loss, or pulse oximetry to detect possible Critical Congenital Heart Disease.

### **4.0 Testing of Newborns**

- 4.1 Dried bloodspot testing:
  - 4.1.1 The health care provider should inform the parent or guardian of the reasons for and the procedures used in the newborn screening tests as well as the potential health consequences of refusing screening. Health care

providers should perform screening tests on newborn infants unless the parent, guardian or custodian of the newborn refuses screening.

4.1.2 The laboratory designated by the Department shall test for the following diseases from the dried bloodspots:

- 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-OH 3-CH<sub>3</sub> glutaric aciduria (HMG)
- Argininosuccinic acidemia (ASA)
- Beta-ketothiolase deficiency (BKT)
- Biotinidase deficiency (BIOT)
- Carnitine uptake defect (CUD)
- Citrullinemia (CIT)
- Congenital adrenal hyperplasia (CAH)
- Congenital hypothyroidism (CH)
- Cystic fibrosis (CF)
- Galactosemia (Classical) (GALT)
- Glutaric acidemia type I (GA I)
- Hb S/Beta-thalassemia (Hb S/BTh)
- Hb S/C disease (Hb S/C)
- Homocystinuria (HCY)
- Isovaleric acidemia (IVA)
- Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD)
- Maple syrup urine disease (MSUD)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Methylmalonic acidemia (Cbl A, B)
- Methylmalonic acidemia (mutase deficiency) (MUT)
- Mucopolysaccharidosis Type I (MPS 1)
- Multiple carboxylase deficiency (MCD)
- Phenylketonuria (PKU)
- Pompe Disease
- Propionic acidemia (PROP)
- Severe combined immunodeficiency (SCID)
- Sickle cell anemia (Hb SS disease) (SS)
- Spinal Muscular Atrophy (SMA)
- Trifunctional protein deficiency (TFP)
- Tyrosinemia type I (TYR I)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
- X-Linked Adrenoleukodystrophy (X-ALD)

- 4.2 Point-of-care testing:
  - 4.2.1 Health care providers should perform a hearing loss screening test on newborn infants unless the parent, guardian or custodian of the newborn refuses screening.
  - 4.2.2 Health care providers shall perform a screening for critical congenital heart disease (CCHD) on every newborn that reflects the standard of care, unless a congenital heart defect was detected prenatally. This screening may include pulse oximetry or other methodologies that reflect the standard of care.

## **5.0 Reporting**

- 5.1 All results of dried bloodspots testing shall be sent by the designated laboratory to the Department for surveillance of chronic diseases.
- 5.2 Health care providers shall provide CCHD screening data to the Department's Newborn Screening Program upon request.
- 5.3 Health care providers may report results of point-of-care newborn screenings to the Department for surveillance of chronic diseases.
- 5.4 The screening health care provider will send documentation of a parent or guardian's refusal for newborn screenings to the Vermont Department of Health, Vermont Newborn Screening Program.

## **6.0 Quality Assurance**

The Department of Health will provide training and technical assistance to hospitals and health care providers in the implementation of the newborn screening program to assure that the program operates according to current standards of practice as established by the American Academy of Pediatrics, the Centers for Disease Control and Prevention, and other such recognized experts in the field of newborn screening.

## **7.0 Retention and Use of Dried Bloodspot Specimens**

- 7.1 After testing is complete, the testing laboratory shall store residual dried bloodspots for one year and then destroy the samples. Storage and destruction shall be done consistent with nationally recognized laboratory standards and applicable federal requirements relating to the disposal of human blood and body fluids.
- 7.2 All existing dried bloodspot samples held by the Department at the time of adoption of this rule will be stored for one year and then destroyed in a manner consistent with applicable federal requirements relating to the disposal of human blood and body fluids.

- 7.3 If the storage environment of any dried bloodspot specimen is found to have deviated from the required conditions, such that the stability of the specimen is likely to have been affected, the testing laboratory shall first notify the Vermont Newborn Screening Program and then shall destroy the dried bloodspot specimen in a manner consistent with applicable federal requirements relating to the disposal of human blood and body fluids.
- 7.4 Dried bloodspot specimens may be destroyed earlier than one year at the written request of the infant's parent(s) or legal guardian(s).
- 7.5 During the one-year period, at the request of the attending physician, and with written consent of the infant's parent(s) or guardian(s), stored dried bloodspot specimens may be retrieved and used for further testing which would assist in the interpretation of screening results or clinical findings for the infant.
- 7.6 Dried bloodspots may be used without parental consent by the testing laboratory only for the purpose of quality assurance and quality control for routine maintenance and function checks.
- 7.7 Dried bloodspots shall not be used for any other purposes without written consent from a parent or guardian.