Newborn Screening Program Rule

1. Authority

1.1. This rule is adopted pursuant to 18 V.S.A. §§ 102, 115 and 991.

2. Purpose and Scope

2.1. 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. 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. 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 declining screening. Health care providers shall perform screening tests on newborn infants unless the parent, guardian or custodian of the newborn declines screening.

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

- 3-Hydroxy-3-methylglutaric aciduria
- 3-Methylcrotonyl-CoA carboxylase deficiency
- Argininosuccinic aciduria
- β-Ketothiolase deficiency
- Biotinidase deficiency
- Carnitine uptake defect/carnitine transport defect
- Citrullinemia, type I
- Classic galactosemia
- Classic phenylketonuria
- Congenital adrenal hyperplasia
- Cystic fibrosis
- Glutaric acidemia type I
- Glycogen storage disease type II (Pompe)
- Guanidinoacetate Methyltransferase Deficiency
- Holocarboxylase synthase deficiency
- Homocystinuria
- Infantile Krabbe disease
- Isovaleric acidemia
- Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency
- Maple syrup urine disease
- Medium-chain acyl-CoA dehydrogenase deficiency
- Methylmalonic acidemia (cobalamin disorders)
- Methylmalonic acidemia (methylmalonyl-CoA mutase)
- Mucopolysaccharidosis type I
- Mucopolysaccharidosis type II
- Primary congenital hypothyroidism
- Propionic acidemia
- Severe combined immunodeficiencies
- S, βeta-thalassemia
- S,C disease
- S,S disease (Sickle cell anemia)

- Spinal muscular atrophy due to homozygous deletion of exon 7 in SMN1
- Trifunctional protein deficiency
- Tyrosinemia, type I
- Very long-chain acyl-CoA dehydrogenase deficiency
- X-Linked adrenoleukodystrophy

4.2. Point-of-care testing:

4.2.1. Health care providers shall perform a hearing screening test on newborn infants unless the parent, guardian or custodian of the newborn declines screening.

4.2.2. Health care providers shall perform on every newborn a screening for critical congenital heart disease (CCHD) that reflects the standard of care unless the parent, guardian or custodian of the newborn declines screening.

5. Reporting

5.1. All results of dried bloodspot testing shall be sent by the designated laboratory to the Department.

5.2. Health care providers shall provide CCHD screening data or newborn dried bloodspot testing follow-up data (e.g., rescreens, diagnostic evaluations, referrals) to the Department upon request.

5.3. Health care providers shall report results of newborn hearing screens, rescreens, and diagnostic evaluations to the Department.

5.4. Providers who implement services for children birth to 3 years of age who are Deaf and Hard of Hearing (e.g., Teachers of the Deaf and Hard of Hearing, Speech and Language Pathologists, Audiologists, Early Intervention Providers) shall report referrals and enrollments into early intervention services to the Department.

5.5. The screening health care provider shall send documentation of a parent or guardian's declination for newborn dried bloodspot testing and hearing screening to the Department.

6. Quality Assurance

6.1. 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, or other such recognized experts in the field of newborn screening.

7. 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 consistent with nationally recognized laboratory standards and applicable federal requirements relating to the disposal of human blood and body fluids.

7.2. 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.

7.3. Dried bloodspot specimens may be destroyed earlier than one year at the written request of the infant's parent(s) or legal guardian(s).

7.4. 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.

7.5. 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.

7.6. Dried bloodspots shall not be used for any other purposes without written consent from a parent or guardian.

Newborn Screening Program Rule

1. Authority

1.1. This rule is adopted pursuant to ~~3 V.S.A. § 3003;~~ 18 V.S.A. §§ 102, 115 and 9915~~5087~~.

2. Purpose and Scope

2.1. 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. 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. 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 declining~~refusing~~ screening. Health care providers ~~should~~ shall perform screening tests on newborn infants unless the parent, guardian or custodian of the newborn ~~refuses~~ declines 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-CH3~~3-Hydroxy-3-methyl-glutaric aciduria ~~(HMG)~~
- 3-Methylcrotonyl-CoA carboxylase deficiency ~~(3MCC)~~
- Argininosuccinic aciduria~~emia~~ ~~(ASA)~~
- β~~Beta~~-K~~k~~etothiolase deficiency ~~(BKT)~~
- Biotinidase deficiency ~~(BIOT)~~
- Carnitine uptake defect/carnitine transport defect ~~(CUD)~~
- Citrullinemia, type I ~~(CIT)~~
- Classic g~~G~~alactosemia ~~(Classical) (GALT)~~
- Classic ~~P~~phenylketonuria ~~(PKU)~~
- Congenital adrenal hyperplasia ~~(CAH)~~
- ~~Congenital hypothyroidism (CH)~~
- Cystic fibrosis ~~(CF)~~
- ~~Galactosemia (Classical) (GALT)~~
- Glutaric acidemia type I ~~(GA I)~~
- Glycogen storage disease type II (Pompe) ~~Disease~~
- Guanidinoacetate Methyltransferase Deficiency
- ~~Multiple~~ Holocarboxylase synthase deficiency ~~(MCD)~~
- ~~Hb S/Beta-thalassemia (Hb S/BTh)~~
- ~~Hb S/C disease (Hb S/C)~~
- Homocystinuria ~~(HCY)~~
- Infantile Krabbe disease
- Isovaleric acidemia ~~(IVA)~~
- Long-chain L-3 -hydroxy~~OH~~ acyl-CoA dehydrogenase deficiency ~~(LCHAD)~~
- Maple syrup urine disease ~~(MSUD)~~
- Medium-chain acyl-CoA dehydrogenase deficiency ~~(MCAD)~~
- Methylmalonic acidemia (cobalamin disorders~~Cbl A, B~~)
- Methylmalonic acidemia (methylmalonyl-CoA mutase ~~deficiency~~) ~~(MUT)~~
- Mucopolysaccharidosis t~~T~~ype I ~~(MPS 1)~~
- Mucopolysaccharidosis type II

- Primary c~~C~~ongenital hypothyroidism ~~(CH)~~
- ~~Multiple carboxylase deficiency (MCD)~~
- ~~Phenylketonuria (PKU)~~
- Propionic acidemia ~~(PROP)~~
- Severe combined immunodeficiencies~~y~~ ~~(SCID)~~
- ~~Hb S, /β~~Beta-thalassemia ~~(Hb S/BTh)~~
- ~~Hb S,/~~C disease ~~(Hb S/C)~~
- S,S disease (Sickle cell anemia) ~~(Hb SS disease) (SS)~~
- Spinal ~~M~~muscular ~~A~~atrophy due to homozygous deletion of exon 7 in SMN1~~(SMA)~~
- Trifunctional protein deficiency ~~(TFP)~~
- Tyrosinemia, type I ~~(TYR I)~~
- Very long-chain acyl-CoA dehydrogenase deficiency ~~(VLCAD)~~
- X-Linked a~~A~~drenoleukodystrophy~~(X ALD)~~

4.2. Point-of-care testing:

4.2.1. Health care providers ~~should~~ shall perform a hearing ~~loss~~ screening test on newborn infants unless the parent, guardian or custodian of the newborn declines~~refuses~~ screening.

4.2.2. Health care providers shall perform on every newborn a screening for critical congenital heart disease (CCHD) ~~on every newborn~~ that reflects the standard of care unless the parent, ~~or~~ guardian or custodian of the newborn declines screening.~~, unless a congenital heart defect was detected prenatally. This screening may include pulse oximetry or other methodologies that reflect the standard of care.~~

5. Reporting

5.1. All results of dried bloodspot~~s~~ 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 or newborn dried bloodspot testing follow-up data (e.g., rescreens, diagnostic evaluations, referrals) to the Department~~'s Newborn Screening Program~~ upon request.

5.3. Health care providers ~~may~~ shall report results of ~~point of care~~ newborn ~~and early periodic~~ hearing screens, rescreens, and diagnostic evaluations ~~ings~~ to the Department ~~for surveillance of chronic diseases~~.

~~5.3.~~5.4. Providers who implement services for children birth to 3 years of age who are Deaf and Hard of Hearing (e.g., Teachers of the Deaf and Hard of Hearing, Speech and Language Pathologists, Audiologists, Early Intervention Providers) shall report referrals and enrollments into early intervention services to the Department.

~~5.4.~~5.5. The screening health care provider ~~will~~ shall send documentation of a parent or guardian's declination~~refusal~~ for newborn dried bloodspot testing and hearing screening~~s~~ to the ~~Vermont~~ Department ~~of Health,, Vermont Newborn Screening Program~~.

6. Quality Assurance

6.1. 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, or~~and~~ other such recognized experts in the field of newborn screening.

7. 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.~~7.2. 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.~~7.3. 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.~~7.4. 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.~~7.5. 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.~~7.6. Dried bloodspots shall not be used for any other purposes without written consent from a parent or guardian.