DEPARTMENT OF HEALTH AND SOCIAL SERVICES
DIVISION OF PUBLIC HEALTH
4100 Family Health Services

4107 Screening of Newborn Infants for Metabolic, Hematologic, Endocrinologic, and Certain Structural Disorders

1.0 Authority and Purpose

Under the authority granted the Department of Health and Social Services, Division of Public Health under 16 **Del.C.** §122(1), 16 **Del.C.** §122(3)(h), 16 **Del.C.** Ch. 8C and 29 **Del.C.** §7904 the Department adopts the following regulations pertaining to the screening of newborns for various disorders.

The purpose of these regulations is to describe the procedures for the Newborn Screening Program where each newborn delivered in the state must be provided a panel of screening tests to identify certain metabolic, hematologic, endocrinologic, and certain structural disorders that may result in developmental delay, cognitive disabilities, serious medical conditions, or death.

The responsibility for implementation of the regulations rests with the institution in which the infant is born. If an infant is born outside a facility, the responsibility for implementation of the regulations rests with the attending delivering physician or midwife, the newborn's primary health care provider and the parent or legal guardian.

16 DE Reg. 1182 (05/01/13)

23 DE Reg. 378 (11/01/19)

2.0 Definitions

"Blood Specimen for Metabolic, Hematologic and Endocrinologic Disorders" means a dried blood spot on a special filter paper utilized for screening (not diagnostic) tests to establish the likely presence of metabolic, hematologic or endocrinologic disorders.

"Certain Structural Disorders" includes critical congenital heart defects and other structural disorders.

"Designated Laboratory" is the laboratory or laboratories, which have been selected by the Division of Public Health to perform these services.

"Endocrinologic Disorder" means the absence or deficiency of a hormone resulting in interference with normal health, growth or development. These disorders include Congenital Hypothyroidism (CH) and Congenital Adrenal Hyperplasia (CAH).

"Hematologic Disorder" means, in these regulations, a condition in which a variation in one or more of the hemoglobin structural genes or in one or more of the genes involved in hemoglobin synthesis produces a variation in hemoglobin structure or synthesis, which results in variation in hemoglobin function. The term "**hemoglobinopathies**" includes sickle cell anemia, sickle cell hemoglobin C disease (SC disease), sickle beta thalassemia, beta thalassemia, alpha thalassemia, hemoglobin C disease and other clinically important variations in hemoglobin structure or synthesis.

"Kit" means any or all parts of the combined materials, laboratory filter paper, specimen forms, Newborn Screening Program brochure, and/or other components provided by the State Newborn Screening Program for the purposes of collection of the blood spot specimen and for submission of the blood spot specimen for laboratory screening.

"Metabolic Disorder" means a disorder caused by a genetic alteration, which results in a defect in the structure or function of a specific enzyme or other protein. These disorders include, but are not limited to, Phenylketonuria (PKU), Galactosemia, Maple Syrup Urine Disease (MSUD), and Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency.

"Newborn Infant" means any infant born in the state who is under 4 weeks of age.

"Pulse Oximetry Screening" is non-evasive test allowing for determination of the oxygen saturation of a patient's hemoglobin to screen for a critical congenital heart defect.

"Satisfactory Specimen" means a blood spot specimen on which an accurate laboratory analysis for the various disorders can be performed.

"The Newborn Screening Advisory Committee" means a committee, established through the Division of Public Health Newborn Screening Program, convened to provide advice and guidance to the Program.

Members are appointed by the Governor and include: three individuals or parents of individuals affected by disorders identified by the screening panel; an ethicist; an attorney not employed by the State; three pediatric physicians; the Medical Director for the Division of Public Health, or their designee; the Laboratory Director for the Division of Public Health, or their designee; a representative from the Department of Services for Children, Youth, and Their Families; the Chair of the Midwifery Advisory Council, or their designee; and a member of the public. The Committee will meet at least semi-annually.

"Unsatisfactory Specimen" means a blood spot specimen that is of insufficient quantity; or a blood spot specimen on which an accurate analysis for the various disorders cannot be performed.

16 DE Reg. 1182 (05/01/13)

23 DE Reg. 378 (11/01/19)

3.0 Determination of Required Screens

The Director of the Division of Public Health or designee shall determine the disorders subject to screening tests.

4.0 Persons Responsible for Submitting Blood Spot Specimens and Pulse Oximetry Results for Screening for Metabolic, Hematologic, Endocrinologic, and Certain Structural Disorders

4.1 The person or facility responsible for assuring that a satisfactory blood spot specimen and pulse oximetry results are submitted for screening newborns for metabolic, hematologic, endocrinologic and certain structural disorders shall be, in order of responsibility:

4.1.1 The hospital, birthing facility or other licensed health care facility in which the newborn is born;

4.1.2 The newborn's primary health care provider; or, if no provider is identified;

4.1.3 The parent or legal guardian.

4.2 In cases of newborns entering a health care facility before 48 hours of age as result of transfer from another facility or of an infant not born in a hospital or other licensed health care facility, the receiving facility shall be responsible for the timely collection of the blood spot specimen and pulse oximetry screening results.

4.3 In cases of newborns not born in a hospital or other licensed health care facility, and not transferred to a health care facility, the timely collection of the blood spot specimen and pulse oximetry screening results shall be the responsibility of the following, in order of responsibility:

4.3.1 The attending delivering physician, or midwife; or in the absence of such a person;

4.3.2 The newborn's primary health care provider; or, if no provider is identified;

4.3.3 The parent or legal guardian.

16 DE Reg. 1182 (05/01/13)

23 DE Reg. 378 (11/01/19)

5.0 Manner of Submitting Blood Spot Specimens and Pulse Oximetry Results

5.1 All dried blood spot specimens submitted to the designated laboratory for testing shall be collected using kits available from the Newborn Screening Program office and/or designated laboratory.

5.2 Blood spot specimens collected for screening shall be forwarded from the collecting facility to the designated laboratory within 24 hours of collection, either by the designated Division of Public Health courier, by mail, or via the service provided by the designated laboratory.

5.3 Pulse oximetry screening results shall be forwarded to the Division of Public Health electronically by the 15th of each month for births occurring in the previous calendar month.

16 DE Reg. 1182 (05/01/13)

23 DE Reg. 378 (11/01/19)

6.0 Timing of Collecting the Blood Spot Specimen and Pulse Oximetry Screening for Screening Infants By Hospitals, Birth Centers or Midwives

6.1 A blood spot specimen for screening for metabolic, hematologic, and endocrinologic disorders shall be collected by the hospital, birth center, or midwife by 72 hours of age regardless of feeding history or medical condition unless the newborn falls into one of the following categories:

6.1.1 For infants born inside or outside of a hospital or other health care facility, or infants born outside of a hospital or other health care facility and transferred to the hospital for continuing care prior to 48 hours of age, the hospital to which the newborn child has been transferred shall collect a specimen from the newborn child, regardless of feeding history or medical condition, as close to arrival as possible.

6.1.2 For pre-term or sick newborns, the initial blood spot specimen shall be collected between 24 and 48 hours after birth. The specimen may be collected prior to 24 hours of age if the infant is receiving blood products. If the first specimen is collected prior to 24 hours of age, a second specimen shall be collected at 8 – 10 days of age or prior to discharge, whichever occurs first. For infants weighing less than 1800 grams at birth, a second specimen shall be collected at 28 days of life or prior to discharge, whichever comes first.

6.1.3 For infants discharged from a hospital or other health care facility before 24 hours of age, a blood spot specimen shall be obtained immediately prior to discharge from the facility and a second dried blood spot specimen shall be obtained after 3 days of age and before 14 days of age.

6.2 Birth facilities or care providers responsible for screening newborns shall adopt protocols consistent with the scientific statement regarding the role of pulse oximetry from the American Heart Association and American Academy of Pediatrics (Pediatrics, 2012: 129 (1) pp. 190-192).

6.3 The data elements to be reported for pulse oximetry screening of newborns to the Division of Public Health are:

6.3.1 The number of births in a birthing facility each month;

6.3.2 The number of pulse oximetry screenings on newborn infants performed each month;

6.3.3 The number of positive and negative screens recorded;

6.3.4 For those infants who do not receive a screen, a reason for not being screened; and

6.3.5 The identity of the infants who fail the screen including their diagnostic evaluation and disposition.

16 DE Reg. 1182 (05/01/13)

23 DE Reg. 378 (11/01/19)

7.0 Procedures for Follow Up of Dried Blood Spot Specimens Designated as Abnormal or Suspicious

7.1 The hospital/facility of birth/midwife to which a newborn is transferred shall develop adequate procedures to ensure a satisfactory blood spot specimen is collected by the time each newborn is two days of age. The sample must be taken from every newborn unless a refusal form signed by the parent or legal guardians is obtained.

7.2 The hospital/facility of birth/midwife to which a newborn is transferred and the primary health care provider of the newborn shall cooperate with the Newborn Screening Program in completing follow up of newborns whose blood spot specimen result is designated as “unacceptable”, “presumptive positive” or “inconclusive.” This cooperation shall include:

7.2.1 Providing appropriate demographic information to the Newborn Screening Program as requested on each baby; and

7.2.2 Providing the Newborn Screening Program with clinical information on each newborn as necessary for interpretation of the results of the screening.

16 DE Reg. 1182 (05/01/13)

23 DE Reg. 378 (11/01/19)

8.0 Reporting of Results of Newborn Screening Tests

8.1 The designated laboratory shall report the results to the Newborn Screening Program as designated in the contract.

8.2 The Newborn Screening Program shall contact with abnormal results the primary health care provider in writing and/or by telephone.

8.3 A copy of the Newborn Screening laboratory report shall be available to the primary health care provider, the birth facility or midwife, and upon request to the parent or legal guardian.

23 DE Reg. 378 (11/01/19)

9.0 Confidentiality of Records

9.1 The Newborn Screening Program shall maintain and treat as confidential all newborn screening communications with facilities, families and health care providers. The Newborn Screening Program shall maintain and treat as confidential a record of every newborn in whom a diagnosis of one or more of the various metabolic, hematologic, or endocrinologic disorders is confirmed.

9.2 Information may be disclosed by the Newborn Screening Program in summary forms, which do not identify individuals. Individuals or institutions requesting summary data must submit a proposal to the Newborn Screening Program and to the Institutional Review Board of the Division of Public Health.

9.3 Dried blood-spots will be retained for a period of three years under appropriate conditions. The stored specimens will only be used for activities to improve the screening program and/or develop new screening tests.

16 DE Reg. 1182 (05/01/13)

10.0 Fees for Newborn Screening Tests Performed in the Designated Laboratory

10.1 The Division of Public Health Newborn Screening Program shall bill the facility or individual for services provided for each newborn screened under these regulations including but not limited to, the cost of the kits for collection of specimens, the laboratory fee for analysis, and administrative costs. The fee will be determined annually (in July) based on cost of the program.

10.2 No Delaware newborn shall be denied testing for hereditary disorders because of inability of the newborn's parent or legal guardian to pay the fee. A "Statement of Fee Exemption" form will be provided to the practitioner or parent requesting exemption from fees. This form must be completed and submitted to the Newborn Screening Program Office within 30 days of birth.

16 DE Reg. 1182 (05/01/13)

23 DE Reg. 378 (11/01/19)

11.0 Exemption from Screening

11.1 A newborn may be excused from screening if the parent or legal guardian objects to the tests for any reason.

11.2 In the event an exemption is claimed from the requirements for screening, the person otherwise responsible for submitting the specimen for screening shall be responsible for submitting a completed refusal form to the Delaware Newborn Screening Program Office, signed by the infant's parent or legal guardian.

11.2.1 The Newborn Screening Refusal Form will be provided through the Newborn Screening Program Office.

16 DE Reg. 1182 (05/01/13)

23 DE Reg. 378 (11/01/19)

12.0 Penalty for Non-compliance

Under the Authority granted to the Department of Health and Social Services, Division of Public Health under 16 **Del.C** §107, "whoever refuses, fails or neglects to perform the duties required under this chapter, or violates, neglects or fails to comply with the duly adopted regulations or orders of the Division shall be fined not less than $100 and not more than $1,000, together with costs, unless otherwise provided by law."

8 DE Reg. 100 (07/01/04)

16 DE Reg. 1182 (05/01/13)

23 DE Reg. 378 (11/01/19)