216-RICR-20-05-1

TITLE 216 – DEPARTMENT OF HEALTH

CHAPTER 20 – COMMUNITY HEALTH

SUBCHAPTER 05 – MATERNAL AND CHILD HEALTH

PART 1 – NEWBORN METABOLIC, ENDOCRINE, HEMOGLOBIN, AND OTHER DISORDERS SCREENING PROGRAM AND NEWBORN HEARING IMPAIRMENT SCREENING PROGRAM

1.1 Authority

A. ~~These rules and regulations are~~This Part is promulgated pursuant to the authority conferred under R.I. Gen. Laws §§ 23-13-9, 13 and 14, and are established for the purpose of updating a comprehensive metabolic, endocrine, hemoglobin, and other disorders ~~y~~ screening program for newborns and a newborn hearing impairment screening program and for adopting a fee structure for said programs.

1.2 Definitions

A. Wherever used in this Part the following terms shall be construed as follows:

1. "Act" means R.I. Gen. Laws Chapter 23-13 entitled, "Maternal and Child Health Services for Children with Special Health Care Needs."

2. “Department” means the Rhode Island Department of Health.

3. "Director" means the Director of the Rhode Island Department of Health or their designee.

4. "Healthcare facilities" means as defined in R.I. Gen. Laws § 23-17-2(9).

5. "Healthcare provider" means any person licensed by the State of Rhode Island to provide healthcare services, including, but not limited to, a physician, healthcare facility, nurse, pharmacist, and any officer, employee or agent of that provider acting in the course and scope of their employment or agency related to or supportive of health services.

6. "Midwife" means a person who has successfully completed an approved educational program in midwifery and is licensed to practice midwifery in Rhode Island pursuant to Midwives (Part 40-05-23 of this Title) and R.I. Gen. Laws § 23-13-9.

7. "Newborn disease" means conditions that have their origin in mutational events that alter the genetic constitution of an individual and/or disrupts normal functions through some other disease mechanism.

8. “Newborn screening program” means the Rhode Island Department of Health, Office of Newborn Screening and Follow-Up program of screening all newborns for metabolic, endocrine, hemoglobin, and other disorders as well as hearing and developmental conditions.

9. "Newborn testing for hearing impairment” means screening and evaluation through the Rhode Island Early Hearing Detection and Intervention Program (RI-EHDI), using procedures prescribed by the Director.

10. "Physician" means as defined in R.I. Gen. Laws R.I. Gen. Laws § 5-37-1(14).

1.3 Newborn Metabolic, Endocrine, Hemoglobin, and Other Disorders Screening Program

A. The physician, midwife, nurse practitioner, or physician assistant attending a newborn child shall cause said child to be subject to screening tests for the conditions listed below. ~~If a physician is not present at the birth of a newborn, a midwife may cause said child to be subject to screening tests for the conditions listed below.~~ Notwithstanding the foregoing, if parents of a newborn child object to the screening tests on the grounds that such tests conflict with their religious tenets and practices, such tests shall not be performed, in accordance with R.I. Gen. Laws § 23-13-14.

1. Metabolic Disorders

   a. Amino Acid Metabolism Disorders

      (1) Argininosuccinic Acidemia

      (2) Citrullinemia

      (3) Homocystinuria

      (4) Maple Syrup Urine Disease

      (5) Phenylketonuria

      (6) Tyrosinemia Type I

   b. Organic Acid Metabolism Disorders

      (1) Beta-Ketothiolase Deficiency

(2) Glutaric Acidemia Type I

(3) Hydroxymethylglutaric aciduria, HMG-CoA lyase Deficiency, or 3-OH 3-CH3 glutaric aciduria

(4) Isovaleric Acidemia

(5) 3-Methylcrotonyl-CoA Carboxylase Deficiency

(6) Methylmalonic Acidemia cblA and cblB forms

(7) Methylmalonic Acidemia due to mutase deficiency

(8) Multiple Carboxylase Deficiency

(9) Propionic Acidemia

c. Fatty Acid Oxidation Disorders

(1) Carnitine Uptake Defect

(2) Long-chain 3-OH acyl COA Dehydrogenase Deficiency (LCHAD)

(3) Medium-chain acyl CoA Dehydrogenase Deficiency (MCHAD)

(4) Very Long-chain acyl CoA Dehydrogenase Deficiency (VLCAD)

(5) Trifunctional Protein Deficiency

2. Endocrine Disorders

a. Congenital Adrenal Hyperplasia

b. Congenital Hypothyroidism

3. Hemoglobin Traits and Disorders

a. Sickle Cell Anemia

b. Hemoglobin S/Beta-Thalassemia

c. Hemoglobin S/C Disease

d. Others detectable through hemoglobin electrophoresis

5. Other Disorders

a. Biotinidase Deficiency

b. Cystic Fibrosis

c. Galactosemia

d. Severe Combined Immunodeficiency (SCID)

e. Critical Congenital Heart Disease

f. Glycogen Storage Disease Type II (Pompe)

g. Spinal Muscular Atrophy (SMA)

h. X-linked Adrenoleukodystrophy (X-ALD)

i. Mucopolysaccharidosis Type I (MPS-I)

j. Mucopolysaccharidosis Type II (MPS-II)

k. Guanidinoacetate Methyltransferase (GAMT) Deficiency

l. Infantile Krabbe Disease

B. The hospital or healthcare facility, or, in the event the birth occurred in a location other than a healthcare facility, the physician, midwife, nurse practitioner, or physician assistant attending a newborn child must cause a filter specimen for the newborn, on a slip supplied by the Department, to be submitted to the laboratory designated by the Department. ~~If a physician is not present at the birth of a newborn, a midwife may cause a filter specimen for the newborn, on a slip supplied by the Department, to be submitted to the laboratory designated by the Department.~~

C. Any laboratory designated by the Department to analyze newborn screening tests must be approved by the Director to perform the tests cited in § 1.3(A) of this Part and as required in this Part.

1. All reports of newborn screening tests must be communicated by the designated laboratory to the Department's designee for follow-up. All such reports shall include actual value and reference ranges used for each disorder.

2. Any reports of inconclusive results must be communicated by the Department's designee for follow-up to the newborn's primary care physician for repeat testing.

3. Any reports of positive results must be communicated by the Department's designee for follow-up to the newborn's primary care physician and any applicable pediatric specialty diagnostic clinic for diagnosis confirmation.

4. Confirmation of diagnosis must be communicated to the Newborn Screening Program by the confirming healthcare provider, and/or specialty diagnostic clinic, within ninety (90) days of confirmation of the diagnosis. Such confirmation must include:

   a. Each confirmed newborn screening diagnosis;

   b. Diagnostic test type;

   c. Treatment type; and

   d. Any additional information the Director may require for surveillance, or for other program or grant purposes.

1.4 Testing for Hearing Impairment

A. Pursuant to R.I. Gen. Laws § 23-13-13, the physician attending a newborn child shall cause said child to be subject to newborn testing for hearing impairment in accordance with procedures approved by the Department. If a physician is not present at the birth of a newborn, a midwife may cause said child to be subject to newborn testing for hearing impairment in accordance with procedures approved by the Department. Notwithstanding the foregoing, no hearing impairment test shall be made as to any newborn infant if the parents of that newborn child object to the test on the grounds that such testing conflicts with their religious tenets and practices, in accordance with R.I. Gen. Laws § 23-13-13(a).

1.5 Fees

A. The fee for the newborn screening (per newborn) and the coordination fee for testing for hearing impairment in newborns (per newborn) shall be as set forth in the Fee Structure for Licensing, Laboratory and Administrative Services Provided by the Department of Health (Part 10-05-2 of this Title).

1.6 Designation of the Newborn Screening Program as a Covered Benefit

A. The newborn screenings and testing for hearing impairment shall be a covered benefit reimbursable by all health insurers, as defined in R.I. Gen. Laws § 27-38.2-2(4), providing health insurance coverage in Rhode Island except for supplemental policies which only provide coverage for specific diseases, hospital indemnity, Medicare supplement or other supplemental policies.

B. When the patient is eligible for Medical Assistance under the provisions of R.I. Gen. Laws Chapter 40-8, Medical Assistance, or R.I. Gen. Laws Chapter 42-12.3, Health Care for Children and Pregnant Women, the cost of the newborn screenings and testing for hearing impairment shall be paid in accordance with R.I. Gen. Laws §§ 23-13-13(c) and 23-13-14(b).

C. Pursuant to R.I. Gen. Laws §§ 23-13-13(c) and 23-13-14(b), in the absence of a third-party payor, the cost for the newborn screenings and testing for hearing impairment shall be paid by the hospital or other healthcare facility where the birth occurred.

D. Any healthcare provider compensated for the newborn screenings and testing for hearing impairment shall reimburse the Department for such newborn screenings and testing for hearing impairment in accordance with Part 10-05-2 of this Title, Fee Structure for Licensing, Laboratory and Administrative Services Provided by the Department of Health.