CABINET FOR HEALTH AND FAMILY SERVICES
Department for Public Health
Division of Laboratory Services
(Amended at ARRS Committee)

902 KAR 4:030. Newborn screening program.

RELATES TO: KRS 194A.050, 211.090, 211.180(1), 214.155
STATUTORY AUTHORITY: KRS 194A.050(1), 211.090(3), 214.155
NECESSITY, FUNCTION, AND CONFORMITY: KRS 214.155 requires the Cabinet for Health and Family Services to operate a newborn screening program for inborn errors of metabolism and other inherited and congenital disorders and conditions, and to establish a schedule of fees to cover the actual costs to the cabinet for the program. This administrative regulation requires that infants be tested for inborn errors of metabolism and other inherited and congenital disorders and conditions as specified in KRS 214.155, and establishes the schedule of fees to cover actual costs of the newborn screening program. The selection of screened conditions is based upon the recommended uniform screening panel as authored by the American College of Medical Genetics and commissioned by the Health Resources and Services Administration, U.S. Department of Health and Human Services.

Section 1. Definitions.
(1) "Blood spot testing" means laboratory testing that is performed on newborn infants to detect a wide variety of inherited and congenital disorders and conditions by using a laboratory-authorized filter paper specimen card.
(2) "Critical congenital heart disease" or "CCHD" means an abnormality in the structure or function of the heart that exists at birth and places an infant at significant risk of disability or death if not diagnosed and treated soon after birth.
(3) "Diagnostic echocardiogram" means a test that uses ultrasound to provide an image of the heart that is performed by a technician trained to perform pediatric echocardiograms.
(4) "Laboratory" means the Division of Laboratory Services within the Cabinet for Health and Family Services, Department for Public Health.
(5) "Pediatric cardiologist" means a pediatrician that is board-certified to provide pediatric cardiology care.
(6) "Program" means the Newborn Screening Program for inherited and congenital disorders and conditions operated by the Cabinet for Health and Family Services, Department for Public Health.
(7) "Pulse oximetry testing" means a noninvasive test that estimates the percentage of hemoglobin in blood that is saturated with oxygen.
(8) "Submitter" means a hospital, primary care provider, health department, birthing center, laboratory, or midwife submitting an infant's blood specimen for the purpose of newborn screening.

Section 2. Tests for inborn errors of metabolism or other inherited or congenital disorders and conditions for newborn infants as part of newborn screening shall be consistent with the U.S. Department of Health and Human Services' Recommended Uniform Screening Panel and include the following:
(1) 2-Methyl-3-hydroxybutyric aciduria (2M3HBA);
(2) 2-Methylbutyryl-CoA dehydrogenase deficiency (2MBDH);
(3) 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC);
(4) 3-Methylglutaconic aciduria (3MGA);
(5) 3-Hydroxy 3-Methylglutaric aciduria (HMG);
(6) Argininemia (ARG);

(7) Argininosuccinic acidemia (ASA);
(8) Beta-ketothiolase deficiency (BKT);
(9) Biotinidase disorder (BIOT);
(10) Carnitine acylcarnitine translocase deficiency (CACT);
(11) Carnitine palmitolytransferase deficiency I (CPT-I);
(12) Carnitine palmitolytransferase deficiency II (CPT-II);
(13) Carnitine uptake defect (CUD);
(14) Citrullinemia type I (CIT-I);
(15) Citrullinemia type II (CIT-II);
(16) Congenital adrenal hyperplasia (CAH);
(17) Congenital hypothyroidism (CH);
(18) Critical congenital heart disease (CCHD);
(19) Cystic fibrosis (CF);
(20) Ethylmalonic encephalopathy (EE);
(21) Galactosemia (GAL);
(22) Glutaric acidemia type I (GA I);
(23) Glutaric acidemia type II (GA-II);
(24) Glycogen storage disease type II (GSD-II, Pompe Disease);
(25) Guanidinoacetate methyltransferase deficiency (GAMT);
(26) Homocystinuria (HCY);
(27) Hypermethioninemia (MET);
(28) Hyperphenylalinemia (H-PHE);
(29) Isobutyryl-CoA dehydrogenase deficiency (IBG);
(30) Isovaleric acidemia (IVA);
(31) Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD);
(32) Malonic academia (MAL);
(33) Maple syrup urine disease (MSUD);
(34) Medium-chain acyl-CoA dehydrogenase deficiency (MCAD);
(35) Methylmalonic acidemia (Cbl A,B);
(36) Methylmalonic acidemia (Cbl C,D);
(37) Methylmalonic acidemia mutase deficiency (MUT);
(38) Mucopolysaccharidosis type I (MPS-I, Hurler's Disease);
(39) Mucopolysaccharidosis type II (MPS-II, Hunter's Disease);
(40) Multiple carboxylase deficiency (MCD);
(41) Non-ketotic Hyperglycinemia (NKHG);
(42) Phenylketonuria (PKU);
(43) Propionic acidemia (PA);
(44) Severe combined immunodeficiency (SCID);
(45) Short-chain acyl-CoA dehydrogenase deficiency (SCAD);
(46) Sickle cell disease (Hb S/S);
(47) Sickle cell hemoglobin C disease (Hb S/C);
(48) Sickle cell S Beta Thalassemia (Hb S/Th);
(49) Spinal muscular atrophy (SMA);
(50) Trifunctional protein deficiency (TFP);
(51) Tyrosinemia type I (TYR-I);
(52) Tyrosinemia type II (TYR-II);
(53) Tyrosinemia type III (TYR-III);
(54) Various Hemoglobinopathies (includes Hb E);
(55) Very long-chain acyl-CoA deficiency (VLCAD); and
(56) X-linked adrenoleukodystrophy (X-ALD).

Section 3. Tests for inborn errors of metabolism or other inherited or congenital disorders and conditions for newborn infants as part of newborn screening shall include the following disorder that is not recommended by the U.S. Department of Health and Human Services, but is required by Kentucky law: Krabbe Disease (KD).

Section 4. Submitter Responsibilities.

(1) Except as provided in KRS 214.155(3) and (5), the administrative officer or other person in charge of the hospital or institution caring for newborn infants and the attending primary care provider or midwife shall administer to, or verify administration of tests to, every infant in its care prior to hospital discharge:

(a) A blood spot test to detect inborn errors of metabolism and other inherited and congenital disorders and conditions identified in Sections 2 and 3 of this administrative regulation; and

(b) Pulse oximetry testing to detect critical congenital heart disease.

(2) If a baby is not born in a hospital or institution, the attending primary care provider or midwife shall ensure that both tests required by subsection (1) of this section are:

(a) Administered between twenty-four (24) and forty-eight (48) hours of age;

(b) Acted upon if abnormal; and

(c) Reported to the program by fax or by the cabinet's web-based system.

(3) A capillary blood spot specimen shall be obtained from a newborn infant not requiring an extended stay due to illness or prematurity between twenty-four (24) and forty-eight (48) hours of age.

(4) If the infant is to remain in the hospital due to illness or prematurity, the hospital shall obtain the capillary blood spot specimen from that infant after twenty-four (24) and before seventy-two (72) hours of age.

(5) Except as provided by subsection (6) of this section, the pulse oximetry testing shall be performed when the infant is twenty-four (24) hours of age or older and shall occur prior to discharge.

(6) If the infant is discharged prior to twenty-four (24) hours of age, the blood spot and pulse oximetry testing shall be performed as close to twenty-four (24) hours of age as possible.

(7) If an infant is transferred from the birth hospital to another hospital during the newborn hospital stay, the requirements established in this subsection shall apply.

(a) The sending hospital shall obtain the capillary blood spot specimen for the newborn screening blood test and the pulse oximetry testing for CCHD if the infant is twenty-four (24) hours of age or more when the infant is transferred to another hospital.

(b) The receiving hospital shall ensure the newborn screening blood spot test and the pulse oximetry testing are performed if the infant is less than twenty-four (24) hours of age when the infant is transferred.

(8) If an infant expires before the newborn screening blood spot test and pulse oximetry test have been performed, the program shall be notified within five (5) calendar days.

(9) If the information on the filter paper specimen card obtained by the submitter and sent to the laboratory is incomplete or inadequate, then the submitter, upon request of the program, shall:

(a) Attempt to locate the infant and obtain a complete and adequate specimen within ten (10) days; and

(b) Report to the program a specimen that is unable to be obtained within ten (10) days.

(10) A submitter that is responsible for the collection of the initial blood spot specimen and pulse oximetry testing for newborn screening shall:

(a) Provide to an infant's parent or guardian educational materials regarding newborn screening and pulse oximetry testing;

(b) Designate a newborn screening coordinator and physician responsible for the coordination of the facility's newborn screening compliance by having a newborn screening protocol;
(c) Notify the program of the name of the individuals designated in paragraph (b) of this subsection each year in January and if the designated individual changes; and
(d) Develop a written protocol for tracking newborn screening compliance, which shall:
1. Be submitted to the program each year in January; and
2. Include, at a minimum:
a. A requirement that the name of the primary care provider that will be attending the infant after birth or discharge or, if known, the primary care provider who will be caring for the infant after discharge, shall be placed on the filter paper specimen card sent with the initial blood spot specimen to the laboratory. If the infant is in the neonatal intensive care unit, the name of the attending neonatologist may be placed on the filter specimen card sent with the initial blood spot specimen to the laboratory;
b. Verification that:
(i) Each infant born at that facility has had a specimen obtained for newborn screening and pulse oximetry testing on or before discharge;
(ii) All information on the specimen card has been thoroughly completed; and
(iii) The specimen has been submitted appropriately;
c. A process to ensure that final results of the pulse oximetry screening are entered into the cabinet's web-based system; and
d. A procedure to assure the hospital or facility that identifies that an infant has not had a specimen obtained for newborn screening and pulse oximetry testing prior to discharge shall:
(i) Notify the program;
(ii) Use every reasonable effort to locate the infant;
(iii) Notify the parent or guardian and the primary care provider immediately; and
(iv) Recommend that the infant present to the hospital or primary care provider immediately for a newborn screening blood spot specimen and pulse oximetry testing.

(11) A hospital or facility shall report each written refusal, in accordance with KRS 214.155(5), to the program within five (5) calendar days.

Section 5. Blood Specimen Collection.

(1) A capillary blood spot specimen required by Section 4 of this administrative regulation shall be obtained by a heel stick.
(2) Blood from the heel stick shall be applied directly to the filter paper specimen card.
(3) All circles shall be saturated completely using a drop of blood per circle on a filter paper specimen card.
(4) The specimen collector shall provide information requested by the laboratory on the filter paper specimen card.
(5) The capillary blood spot specimen shall be air dried for three (3) hours and then shall be mailed or sent to the laboratory:
(a) Within twenty-four (24) hours of collection of the specimen; or
(b) The next business day in which mail or delivery service is available.
(6) A submitter sending a blood spot specimen via regular mail services shall send the specimen to the following address: Cabinet for Health and Family Services, Department for Public Health, Division of Laboratory Services, 100 Sower Boulevard, Frankfort, Kentucky 40602.

(7) A submitter sending a blood spot specimen via expedited mail services shall ensure the specimen is sent to the following address: Cabinet for Health and Family Services, Department for Public Health, Division of Laboratory Services, 100 Sower Boulevard, Suite 204, Frankfort, Kentucky 40602.

(8) Specimens processed or tracked under the newborn screening program shall be limited to specimens on infants less than six (6) months of age.

Section 6. Unsatisfactory or Inadequate Blood Specimen.

(1) If a specimen is unsatisfactory or inadequate to produce a valid result, the laboratory shall notify the submitter and the parent on the filter paper specimen card that the newborn screen needs to be repeated as soon as possible.

(2) If a requested repeat specimen has not been received within ten (10) business days from the date the repeat request was issued, the program shall notify the parent by mail of the need for a repeat screening test.

Section 7. Special Circumstances - Blood Transfusion. If a newborn infant requires a blood transfusion, the requirements for newborn screening established in this section shall apply.

(1) The hospital shall obtain a capillary blood spot specimen for newborn screening prior to the infant being transfused, except in an emergency situation.

(2) If the pre-transfusion blood spot specimen was obtained before twenty-four (24) hours of age, or if it was not obtained due to an emergency situation, then the hospital or primary care provider shall use all reasonable efforts to obtain a repeat capillary blood specimen from the transfused infant and submit it to the laboratory according to the following schedule:

(a) Seventy-two (72) hours after the last blood transfusion, rescreen for inborn errors of metabolism and inherited and congenital disorders and conditions listed in Sections 2 and 3 of this administrative regulation; and

(b) Ninety (90) days after the last blood transfusion, rescreen for any disorder that relies on red blood cell analysis such as hemoglobinopathies, galactosemia, and biotinidase deficiency.

Section 8. Reporting Results of Newborn Screening Blood Tests.

(1) Normal Results. Upon receipt of a normal lab result, the laboratory shall send the result to the primary care provider and the submitter.

(2) Abnormal Results.

(a) The laboratory shall report abnormal, presumptive positive, or equivocal results of tests for inborn errors of metabolism, inherited and congenital disorders and conditions to the program.

(b) The submitter and primary care provider shall receive a copy of all abnormal, presumptive positive, and equivocal results.

(c) In addition, a primary care provider shall be notified of an abnormal, presumptive positive, or equivocal result in the manner established in this paragraph.

1. Upon receipt of an abnormal, equivocal, or a presumptive positive lab result, the laboratory shall notify the primary care provider listed on the filter paper specimen card within two (2) business days of the result and the need for follow-up testing.

2. Upon receipt of a presumptive positive lab result, the program shall notify the primary care provider listed on the filter paper specimen card of the result and recommend immediate consultation with a university pediatric specialist.

3. If the program is unable to determine the infant's primary care provider to notify them of an abnormal, presumptive positive, or equivocal result and the need for follow-up, the program shall use every available means to notify the infant's parent.

(d) The Cabinet for Health and Family Services shall share pertinent test results with a state university-based specialty clinic or primary care provider who informs the cabinet

they are treating the infant who received the test.

(e) The cabinet may share pertinent test results with the local health department in the infant's county of residence that conducts newborn screening follow-up activities.

(f) A specialty clinic or primary care provider shall report results of diagnostic testing to the program within thirty (30) days or earlier upon request.

(g) If a requested repeat specimen has not been received within ten (10) business days from the date the repeat request was issued, the program shall notify the parent by mail of the need for a repeat screening test.

Section 9. Pulse Oximetry Screening for Critical Congenital Heart Disease. Pulse oximetry screening for critical congenital heart defects required by Section 2 of this administrative regulation shall be consistent with the standard of care according to national recommendations by the American Academy of Pediatrics.

Section 10. Pulse Oximetry Screening Process.

(1) Except as provided by KRS 214.155(3) and subsections (2) and (4) of this section, pulse oximetry testing shall be performed when the infant is between twenty-four (24) and forty-eight (48) hours of age and shall occur no later than the day of discharge.

(2) If the infant is discharged prior to twenty-four (24) hours of age, the blood spot and pulse oximetry testing shall be performed as close to twenty-four (24) hours of age as possible.

(3) An infant in a neonatal intensive care unit shall be screened when medically appropriate after twenty-four (24) hours of age but prior to discharge.

(4) An infant who has been identified with critical congenital heart disease prior to birth or prior to twenty-four (24) hours of age shall be exempt from the pulse oximetry screening process.

(5) Pulse oximetry screening shall be performed by placing pediatric pulse oximetry sensors simultaneously on the infant's right hand and either foot to obtain oxygen saturation results.

(6) If using a single pediatric pulse oximetry sensor, pulse oximetry screening shall be performed on the infant's right hand and either foot, one after the other, to obtain oxygen saturation results.

Section 11. Pulse Oximetry Testing Results.

(1) A passed result shall not require further action if:

(a) The pulse oximetry reading in both extremities is greater than or equal to ninety-five (95) percent; and

(b) The difference between the readings of both the upper and lower extremity is less than or equal to three (3) percent.

(2)

(a) A pending result shall:

1. Occur if:

a. The pulse oximetry reading is between ninety (90) and ninety-four (94) percent; or

b. The difference between the readings of both the upper and lower extremity is greater than three (3) percent; and

2. Be repeated using the pulse oximetry screening in one (1) hour.

(b) If a repeated pulse oximetry screen is also interpreted as pending, it shall be performed again in one (1) hour.

(c) If the pulse oximetry result on the third screen continues to meet the criteria as pending after three (3) screenings have been performed, it shall be considered failed and the procedures established in subsection (3) of this section shall be followed.

(3) A failed result shall:

(a) Occur if:

1. The initial pulse oximetry reading is less than ninety (90) percent in the upper or lower extremity; or

2. The provisions of subsection (2)(c) of this section apply; and

(b) Require the following actions:

1. The primary care provider shall be notified immediately;

2. The infant shall be evaluated for the cause of the low saturation reading; and

3. If CCHD cannot be ruled out as the cause of the low saturation reading, the attending physician or advanced practice registered nurse shall:

a. Order a diagnostic echocardiogram to be performed without delay;

b. Ensure the diagnostic echocardiogram be interpreted as soon as possible; and

c. If the diagnostic echocardiogram results are abnormal, obtain a consultation with a pediatric cardiologist prior to hospital discharge.

Section 12. Reporting Results of Pulse Oximetry Screening.

(1) Final results of the pulse oximetry screening shall be entered into the cabinet's web-based system.

(2) A failed result shall be immediately reported to the program by fax or by the cabinet's web-based system.

Section 13. Newborn Screening Fees.

(1)

(a) A submitter, other than a midwife, obtaining and sending a blood spot specimen to the laboratory shall be billed a fee of $200 for the initial newborn screening test.

(b) A midwife obtaining and sending a blood spot specimen to the laboratory shall be billed a fee of fifty (50) dollars for the initial newborn screening test.

(2) A submitter obtaining and sending a repeat blood spot specimen to the laboratory shall not be charged an additional fee.

(3) Fees due the Cabinet for Health and Family Services shall be collected through a monthly billing system.

(MCH-3; 1 Ky.R. 640; eff. 4-9-1975; Am. 6 Ky.R. 314; eff. 1-2-1980; 9 Ky.R. 386; 555; eff. 10-6-1982; 11 Ky.R. 652; eff. 11-13-1984; 13 Ky.R. 786; eff. 11-11-1986; 14 Ky.R. 2069; eff. 6-22-1988; 21 Ky.R. 578; eff. 9-21-1994; 32 Ky.R. 2036; 1487; 2274; eff. 6-21-2006; 40 Ky.R. 1484; 2449; eff. 6-6-2014; 41 Ky.R. 1222; eff. 4-3-2015; 42 Ky.R. 1650; eff. 3-4-2016; 45 Ky.R. 3553; eff. 8-19-2019; 47 Ky.R. 209; eff. 12-15-2020; 48 Ky.R. 3071; eff. 9-28-2022; 51 Ky.R. 138, 668; eff. 10-23-2024.)

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