NEWBORN SCREENING RULE

LOUISIANA ADMINISTRATIVE CODE

TITLE 48

PUBLIC HEALTH-GENERAL

PART V. Preventive Health Services

Subpart 18. Disability Prevention Program

Chapter 63. Newborn Heel Stick Screening

§6303. Purpose, Scope, Methodology

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§6303. Purpose, Scope, and Laboratory Testing Methodology

A. R.S. 40:1081.1 and 1081.2 requires physicians to test Louisiana newborns for the disorders listed below along with the abbreviations used by the American College of Medical Genetics (ACMG).

1. Disorders of amino acid metabolism:
   a. phenylketonuria (PKU);
   b. maple syrup urine disease (MSUD);
   c. homocystinuria (HCY
   d. citrullinemia, type I (CIT);
   e. argininosuccinate acidemia (ASA); and
   f. tyrosinemia, type I (TYR I).
2. Disorders of fatty acid metabolism:
   a. medium-chain acyl-CoA dehydrogenase deficiency (MCAD);
   b. trifunctional protein deficiency (TFP);
   c. very long-chain acyl-CoA dehydrogenase deficiency (VLCAD);
   d. carnitine uptake defect (CUD); and
   e. long chain-3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD).
3. Disorders of organic acid metabolism:
   a. isovaleric acidemia (IVA);
   b. methylmalonic acidemia (methylmalonyl-CoA mutase, MUT),(cobalamin disorders, CBL A, B);
   c. glutaric acidemia type 1 (GA1);
   d. propionic acidemia (PROP);
   e. 3-hydroxy-3-methylglutaryl–CoA lyase deficiency (HMG);
   f. multiple carboxylase deficiency (MCD) including, but not limited to, holocarboxylase synthetase deficiency;
   g. beta-ketothiolase deficiency (BKT); and

h. 3-methylcrotonyl CoA carboxylase deficiency (3-MCC).

4. Other metabolic disorders:

a. biotinidase deficiency (BIOT); and

b. classic galactosemia (GALT).

5. Endocrine disorders:

a. congenital hypothyroidism (CH); and

b. congenital adrenal hyperplasia (CAH).

6. Hemoglobinopathies (sickle cell diseases):

a. hemoglobin S,S disease (sickle cell anemia) (Hb SS);

b. hemoglobin S,C disease (Hb SC);

c. hemoglobin S, beta-thalassemia disease (Hb S/βTH); and

d. other sickling diseases.

7. Pulmonary disorders:

a. cystic fibrosis (CF).

8. Immune Disorders:

a. severe combined immunodeficiency (SCID).

B. Methodology

1. Filter Paper Specimen Form (Lab10), used in blood specimen collection for neonatal screening, can be obtained from the Genetic Diseases Program by calling 504-568-8254. There are two different types of Lab-10 forms which are color-coded.

a. For patients covered by Medicaid or Managed Care Plans, blue border Lab-10 forms are used. There is no charge to private providers for these blue border forms. The patient's Medicaid number (or mother's number, if the patient has not been issued one) shall be indicated on the form.

b. For private and non-Medicaid patients, red border Lab-10 forms are used. These red border Lab-10 forms are $30 each. The name of the insurance company and policy number shall be included on the form.

2. Private providers should order a mix of red and blue Lab-10 forms from the Genetic Diseases Program to match the Medicaid/non-Medicaid composition of newborns to be screened at their facility. The Lab-10 forms shall be completely filled out.

3. For non-Medicaid patients with a financial status of greater than 100 percent of the poverty guidelines as established by the Louisiana Department of Health (LDH) and who attend a parish health unit for just the newborn screening service, the parent or guardian shall be charged $30 upon registering at the parish health unit.

4. To ensure that specimens for testing are received within 2 to 3 days by the laboratory approved by the Office of Public Health (OPH) to perform newborn screening pursuant to the requirements of this Chapter, all such laboratories shall provide mailing envelopes to submitting hospitals which guarantee a delivery time no longer than 3 days from mailing. An example of an acceptable minimum option would be the use of the United States Postal Service's Flat Rate Priority Mailing Envelopes. The use of all other companies and courier services providing the required level of service stated herein are acceptable.

C. Policy for Pre-Discharge, Repeat Screening and Education to Parents on Repeat Screening

1. Pre-Discharge Screening. All hospitals that have maternity units shall institute and maintain a policy of screening all newborns before discharge regardless of their length of stay in the hospital. The initial screen should occur at greater than 24 hours of birth but shall occur no later than 7 days after birth.

2. Repeat Screening for Specimens Collected before 24 Hours. There is a greater risk of false negative results for specimens collected from babies younger than 24 hours of age. Therefore, full-term, healthy newborns screened prior to 24 hours of age must be rescreened at the first medical visit, preferably between 1 and 2 weeks of age, but no later than the third week of life. Repeat screening should be arranged by the primary pediatrician; however, it may be done by any primary healthcare provider or clinical facility qualified to perform newborn screening specimen collection. For preterm, low birth weight, and sick infants admitted to the neonatal intensive care unit (NICU), an initial specimen should be collected upon admission, a second specimen shall be collected at 48-72 hours after admission and a final specimen shall be collected at 28 days or upon discharge, whichever comes first.

3. Education to Parents on Repeat Screening. To ensure that newborns who need rescreening actually receive the repeat test, hospitals with maternity units must establish a system for disseminating information to parents about the importance of rescreening. This includes infants with an initial unsatisfactory specimen, infants with an initial collection performed at less than 24 hours of age, and infants admitted to the NICU.

D. Notification of Screening Results

1. The Genetic Diseases Program follow-up staff shall notify the appropriate medical provider of the positive screening result by telephone. Otherwise, submitters should receive test results from the State Public Health Laboratory within 5 days after collection. Test results are available to submitters 24 hours a day, 365 days a year through the web-based Secure Remote Viewer (SRV) which is accessed via computer. Information on signing up for and using the SRV can be obtained by calling the Genetic Diseases Program Office at (504) 568-8254. If test results are not available, medical providers may fax in their requests to the following numbers: (225) 219-4905 (Public Health Biochemistry Laboratory) or (504) 568-8253 (Genetics Office). In order to retrieve test results from the SRV, the provider must have the infant’s date of birth plus one of the following: mother’s first name, mother’s last name, baby’s first name or baby’s last name. Test results can also be found by the infant’s medical record number or by the Lab 10 form number.

E. Unsatisfactory Specimens. The accuracy of a test depends on proper collection of the blood spot. Specimens of unsatisfactory quality for testing shall be indicated on the test result slip. Training on collecting adequate specimens can be arranged by calling the Genetics Diseases Program at telephone number (504) 568-8254.

F. Medical/Nutritional Management

1. In order for a patient with PKU or other rare inborn errors of metabolism to receive the special formulas for the treatment of these disorders from the state's Genetic Diseases Program and/or Special Supplemental Nutrition Program for Infants, Women, and Children (WIC), the following guidelines shall be met:

a. The patient shall be a resident of the State of Louisiana.

b. The patient shall receive clinical and dietary management services through a metabolic center to include a medical evaluation at least once annually by a physician who is board certified in biochemical genetics or a medical geneticist physician with written documentation of a medical evaluation and continuing consultation with a physician board certified in biochemical genetics. A licensed registered dietitian must also be on staff and be readily available for both acute and chronic dietary needs of the patient. Children less than 1 year of age shall be seen by the dietitian and medical geneticist at least twice a year. Children greater than 1 year of age shall be seen at least once per year by the dietitian and medical geneticist.

c. The patient shall provide necessary blood specimens for laboratory testing as requested by the treating physician meeting the above requirements. Laboratory test result values for phenylalanine and tyrosine shall be submitted to the Genetics Program Office by the treating medical center within 15 working days after data reduction and interpretation.

d. The patient shall include dietary records with the submission of each blood specimen.

e. All insurance forms relative to charges for special formula shall be signed and submitted by the parent or appropriate family member.

f. The parent or guardian shall inform the Genetics Program Office immediately of any changes in insurance coverage.

g. If a patient fails to comply with these requirements, he/she shall not be able to receive metabolic formula, medications and medical services through the Office of Public Health.

G. Acceptable Newborn Screening Testing Methodologies and Procedures for Medical Providers Not Using the State Laboratory. Laboratories performing or intending to perform the state mandated newborn screening battery on specimens collected on Louisiana newborns shall meet the conditions specified below pursuant to R.S. 40:1081.2.

1. The testing battery shall include testing for the disorders listed in Subsection A above.

2. The laboratory shall perform the newborn screening testing battery on at least 50,000 specimens a year unless the said laboratory has been routinely performing the full screening battery since January 1, 1995.

3. A laboratory shall perform the complete battery at one site. Using two laboratories for completion of the total battery is unacceptable as this increases the risk of error and delay in reporting.

4. When using dried blood spots, only specimen forms using filter paper approved by the Centers for Disease Control and Prevention (CDC) are acceptable.

5. Only the following testing methodologies listed in Table 6303.G.5 are acceptable without prior written approval from the Genetic Diseases Program.

Table 6303.G.5

Disease	Testing Methodology
Disorders of Amino Acid Metabolism Disorders of Fatty Acid Metabolism Disorders of Organic Acid Metabolism (Specific disorders include those as listed under Subsection A)	Tandem Mass Spectrometry (MS/MS)
Biotinidase Deficiency	Time-Resolved Immunofluorescence assay Qualitative or Quantitative Enzymatic Colorimetric or Fluorometric
Galactosemia	Galt enzyme assay Total Galactose

Table 6303.G.5	
Disease	**Testing Methodology**
Hemoglobinopathies (Sickle Cell Diseases)	Cellulose acetate/citrate agar Capillary isoelectric focusing (CIEF) Gel isoelectric focusing (IEF) High Pressure Liquid Chromatography (HPLC) DNA Mutational Analysis Sickle Dex – is NOT Acceptable Controls must include: F, A, S, C, D, E If controls for hemoglobins D and E are not included in the first tier testing methodology, then the second tier testing must be able to identify the presence of these hemoglobins. Result Reporting: by phenotype Positive/negative is NOT acceptable
Congenital Hypothyroidism	Radioimmunoassay (RIA), Fluorescent Immunoassay (FIA) time resolved fluoroimmunoassay, Enzyme Immunoassay (EIA) methods for T4 and/or Thyroid Stimulating Hormone (TSH) which have been calibrated for neonates
Congenital Adrenal Hyperplasia	17 hydroxyprogesterone (17OHP), time resolved fluoroimmunoassay

Table 6303.G.5	
Disease	**Testing Methodology**
Cystic Fibrosis	Primary: Immunoreactive Trypsinogen; Time-Resolved fluoroimmunoassay Second Tier: Deoxyribonucleic Acid (DNA) mutation analysis Qualitative Sweat Conductivity Test is NOT acceptable as a primary screening methodology. Confirmatory Test Methodologies: Quantitative Pilocarpine Iontophoresis Sweat Chloride Test Qualitative Sweat Conductivity Test is NOT recommended.
Severe Combined Immunodeficiencies (SCID)	Real Time Quantitative Polymerase Chain Reaction (RTQPCR)

a. Alternative Methodologies not listed in Table 6303.G.5. New Food and Drug Administration (FDA)-approved methodologies may be used if first found to be acceptable by the Genetics Diseases Program. Approval shall be requested from the Genetic Diseases Program in writing 60 days before the intended date of implementation by mailing the request to:

LDH OPH Genetic Diseases Program
1450 Poydras Street, Suite 2046
New Orleans, Louisiana 70112

b. Approval Process. Requests for approvals of methodologies not listed in Table 6303.G.5 shall be based on documentation of FDA-approved methodologies or on documentation of OPH Laboratory-developed test methodologies, as well as an in-house OPH Laboratory validation study of the applicable methodology proposed for use.

6. The laboratory shall comply with the regulations for proficiency testing as mandated in the Clinical Laboratory Improvement Amendments of 1988 (CLIA 88 §493.1707). When using dried blood spots, the laboratory must participate in a proficiency testing program. The laboratory must report all proficiency testing results to the Genetic Diseases Program Office within one month of receiving the report from the proficiency testing provider.

7. The laboratory shall be able to provide test result data to physicians and nurses on their specific patients by telephone and by FAX or by use of the internet, 24 hours a day 365 days a year.

8. Mandatory Reporting of Positive Test Results Indicating Disease

a. To ensure appropriate and timely follow-up, positive results shall be reported, along with patient demographic information as specified below to the Genetic Diseases Program Office by fax at (504) 568-8253. Receipt of faxed results shall be verified by calling the Genetics Office at (504) 568-8254.

b. Described below are specific time deadlines after data reduction and interpretation for reporting positive results indicating probable disease to the Genetics Diseases Program Office. Laboratories shall make arrangements with the Genetics Diseases Program Office for reporting after hours, weekends and

holidays for positive test results from tandem mass spectrometry and the assays for galactosemia, and congenital adrenal hyperplasia. Notification of presumptive positive results for biotinidase deficiency, sickle cell disease, congenital hypothyroidism and cystic fibrosis shall be made at the beginning of the next business day:

i. metabolic disorders identified by tandem mass spectrometry and for galactosemia—report results within 2 hours;

ii. biotinidase deficiency—report results within 24 hours;

iii. sickle cell disease—report results of FS, FSC, FSA from initial specimens within 24 hours;

iv. congenital hypothyroidism—report within 24 hours;

v. congenital adrenal hyperplasia—report within 2 hours; and

vi. cystic fibrosis—report within 24 hours.

c. The specified information to be reported:

i. - xiii. …

xiv. transfusion given?

Yes ____ No ____

If yes, date of last transfusion (if available): _____

9. Provision of Follow-up Services. To ensure that reporting time deadlines specified under Subparagraph b of Paragraph 8 of this Subsection are met for every positive test result indicating probable disease, a follow-up system must be in operation. The protocol for a follow-up system may rely on the submitting hospital for the follow-up action which must include the following.

a. Locate the infant and ensure diagnostic and medical care:

i. telephone call to medical provider within 24 hours of positive lab result;

ii. if there is no medical provider available, a telephone call should be made to parent/guardian;

iii. if the parent/guardian does not have a telephone, then notify them by certified and regular mail;

iv. if there is no response to mail within five days, a home visit should be made; and,

v. report to the Genetic Diseases Program Office all patients with suspect results who are unable to be located.

b. Results of repeat testing should be obtained.

i. If results are normal, the case can be closed.

ii. If results are abnormal, the case must be reported to the Genetic Diseases Program Office.

10. Reporting requirements of private laboratories to the Genetic Diseases Program Office for public health surveillance and quality assurance purposes.

a. The laboratory shall submit quarterly statistical reports to the Genetic Diseases Program Office that indicate the number of specimens screened by method, the number of specimens unsatisfactory for testing, the number normal and positive, and for screening of hemoglobinopathies, the number by phenotype [see the Genetics Diseases Program Office's address near the end of the Diseases/Testing Methodology table (which may be found under Paragraph 5 of this Subsection)].

b. The laboratory shall electronically report newborn screening results on all Louisiana newborns screened to the Genetic Diseases Program Office on a monthly basis. The file format and data layout shall be determined by the Genetic Diseases Program. Essential patient data is the following and is required to be reported unless "optional" is indicated:

i child's name;

ii. child's last name;

iii. mother's first name;

iv. mother's last name;

v. mother's maiden name (optional);

vi. child's street address;

vii. child's city;

viii. child's state;

ix. child's zip code;

x. child's parish (optional);

xi. child's date of birth (format: mm/dd/yyyy);

xii. child's sex;

xiii. child's race (format: (W)hite, (B)lack, Native American, Asian, other, Hispanic);

xiv. mother's Social Security number (format: 999-99-9999); and

xv. child's test results.

11. The laboratory shall register by letter with the OPH's Genetic Diseases Program each year. This letter shall contain the following and shall be received in the Genetic Diseases Program Office by February 1 each year:

a. assurance of compliance with the requirements described in Subsection G. - G.9. of this Subsection;

b. the type of testing methodologies used;

c. the number of specimens projected to be tested or actually tested annually;

d. the type of specimen(s) used, i.e., filter paper or whole blood; and

e. reporting format for positive/abnormal test results.

H. The Newborn Heel Stick Screening Policy for Result Reporting and Repeat Screening Post Transfusion

1. Whenever possible, a specimen should be collected prior to transfusion.

2. Repeat testing recommended: 3 days after transfusion and 90 days after last transfusion.

3. If the specimen was not collected before transfusion, the laboratory reporting the results to the submitter shall indicate that transfusion may alter all newborn screening results and include the above times for repeat screening.

AUTHORITY NOTE: Promulgated in accordance with R.S. 40:1018.1 and 1081.2

HISTORICAL NOTE: Promulgated by the Department of Health and Human Resources, Office of Preventive and Public Health Services, LR 13:246 (April 1987), amended by the Department of Health and Hospitals, Office of Public Health, LR 17:378 (April 1991), LR 18:1131 (October 1992), LR 20:1386 (December 1994), LR 23:301 (March 1997), LR 27:545 (April 2001), LR 29:1490 (August 2003), LR 32:248 (February 2006), LR 34:442 (March 2008), amended by the Department of Health, Office of Public Health, LR 44:1908 (October 2018).