NEWBORN SCREENING RULE

LOUISIANA ADMINISTRATIVE CODE

TITLE 48

PUBLIC HEALTH - GENERAL

Part V. Public Health Services

Subpart 19. Genetic Diseases Services

Chapter 63.Neonatal Screening

§6303. Purpose, Scope, Methodology

Chapter 63. Newborn Heel Stick Screening

§6303. Purpose, Scope Methodology

A. R.S. 40:1299.1.2.3, 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 (CIT);
   e. Argininosuccinic Aciduria (ASA);
   f. Tyrosinemia type I (TYR I).
2. Disorders of fatty acid metabolism:
   a. Medium Chain Acyl-CoA dehydrogenase Deficiencey (MCAD);
   b. Trifunctional protein deficiency (TFP);
   c. Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD);
   d. Carnitine Uptake Defect (CUD);
   e. Long Chain-3-Hydroxy Acyl-CoA Dehydrogenase Deficiency (LCHAD).
3. Disorders of organic acid metabolism:
   a. Isovaleric Acidemia (IVA);
   b. Methylmalonic Acidemia (MUT),(CBL A, B);
   c. Glutaric Acidemia Type 1 (GA1);
   d. Proprionic Aciduria (PROP);
   e. 3-Hydroxy-3-Methylglutaryl–CoA Lyase (HMG);
   f. Multiple Carboxylase Deficiency (MCD);
   g. β-Ketothiolase Deficiency (BKT);
   h. 3-Methylcrotonyl CoA Carboxylase Deficiency (3MCC).
4. Other metabolic disorders:
   a. Biotinidase Deficiency (BIOT);
   b. Galactosemia (GALT).
5. Endocrine disorders:
   a. Congenital Hypothyroidism (CH);
   b. Congenital Adrenal Hyperplasia (CAH).
6. Hemoglobinopathies (Sickle Cell diseases):
   a. SS disease (Sickle Cell Anemia) (Hb SS);
   b. SC disease (Hb SC);
   c. S/Beta Thalassemia (Hb S/βTH);
   d. Other sickling diseases.

7. Pulmonary disorders:
   a. Cystic Fibrosis (CF).

B. Methodology

1. Filter Paper Specimen Form, (Lab-10) used in blood specimen collection for neonatal screening, can be obtained at parish health units. There are two different types of Lab-10 forms which are color coded.

   a. For patients covered by Medicaid, including those in the Kid-Med Program, blue border forms are used. There is no charge to private providers for these blue border forms.

   b. For private and non-Medicaid patients, red border Lab-10 forms are used. These red border Lab-10 forms are $30.00 each.

2. Private providers should order a mix of red and blue Lab-10 forms from their local parish health unit (or OPH Regional Office for certain areas) to match the Medicaid/non-Medicaid composition of newborns to be screened at their facility. The Lab-10 forms must be completely filled out.

3. For non-Medicaid patients with a financial status of greater than 200% of the Poverty Guidelines as established by the Department of Health and Human Services (DHHS) and who attend a parish health unit for just the newborn screening service, the parent or guardian will be charged $30.00 upon registering at the parish health unit.

4. To ensure that specimens for testing are received within two to three days by the laboratory approved by OPH to perform newborn screening pursuant to the pertaining requirements of this chapter, all such laboratories must provide mailing envelopes to submitting hospitals which guarantee a delivery time no longer than three 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 this service 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. Newborns remaining in the hospital for an extended period should be screened initially no later than seven 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, newborns screened prior to 24 hours of age must be rescreened at the first medical visit, preferably between one and two 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.

3. Education to Parents on Repeat Screening. To ensure that neonates who need rescreening (due to an initial unsatisfactory specimen, an initial collection performed on a baby less than 24 hours old) actually receive the repeat test, hospitals with maternity units must establish a system for disseminating information to parents about the importance of rescreening.

D. Notification of Screening Results

1. The Genetic Diseases Program Follow-up staff notify the appropriate medical provider of the positive screening result by telephone. Otherwise, submitters should receive the result slip from the State Public Health Laboratory within two weeks. Results are also available to submitters 24 hours a day, 365 days a year through the Voice Response System with FAX (VRS) which is accessed by using a touch tone telephone. Information on using VRS can be obtained by calling the Genetic Diseases Program Office at (504) 219-4413. If results are not available, medical providers may fax in their requests to the following numbers: (504) 219-4694 (Public Health Biochemistry Laboratory) or (504) 219-4452 (Genetics Office). To assist the pediatrician's office in the retrieval of the results on the initial specimen of the infant at the first medical visit, the phlebotomist or nurse collecting the initial specimen should tear off the blue carbon of the Lab-10 form and give this to the parent or guardian. The parent should be instructed to bring this copy to the first medical visit.

E. Unsatisfactory Specimens

The accuracy of a test depends on proper collection of the blood spot. Specimens of unsatisfactory quality for testing will be indicated on the result slip. Training on collecting adequate specimens can be arranged by calling the Genetics Nurse at telephone number 504-219-4413.

F. Medical/Nutritional Management

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

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

b. The patient must receive a medical evaluation at least once annually through a metabolic center to include a medical evaluation 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 one year of age must be seen by the dietitian and medical geneticist at least twice a year. Children greater than one year of age must be seen at least once per year by the dietitian and medical geneticist.

c. The patient must 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 must be submitted to the Genetics Program Office by the treating medical center within 15 working days after data reduction and interpretation.

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

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

f. The parent or appropriate family member must inform the Genetics Program office immediately of any changes in insurance.

g. If a patient fails to comply with these requirements, he/she will 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 must meet the conditions specified below pursuant to R.S. 40:1299.1:

1. The testing battery must include testing for disorders listed in Subpart A above.

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

3. A laboratory must 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 (CDC) are acceptable.

5. Only the following testing methodologies are acceptable without prior approval:

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 part A)	Tandem Mass Spectrometry (MS/MS)
Biotinidase Deficiency	Qualitative or Quantitative Enzymatic Colorimetric or Fluorometric
Galactosemia	Galt enzyme assay Total Galactose

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 - NOT Acceptable Controls must include: F, A, S, C, D, E If controls for hemoglobins D and E are not included in the 1st tier testing methodology, then the 2nd 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), Enzyme Immunoassay (EIA) methods for T4 and/or Thyroid Stimulating Hormone (TSH) which have been calibrated for neonates
Congenital Adrenal Hyperplasia	17 hydroxyprogesterone (17OHP)

Disease	Testing Methodology
Cystic Fibrosis	Primary: Immunoreactive Trypsinogen; Second Tier: DNA 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.
New Food and Drug Administration approved methodologies may be used if found to be acceptable by the Genetic Diseases Program. Approval should be requested in writing 60 days before the intended date of implementation (see Genetic Diseases Program mailing address below). Requests for approvals will be based on documentation of FDA approval and an in-house validation study of said methodology.	

6. The laboratory must comply with the regulations for proficiency testing as mandated in the Clinical Laboratory Improvement Amendments of 1988 (CLIA 88 Section §493.1707). When using dried blood spots, the laboratory must participate in the proficiency testing program of the Centers for Disease Control. 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 must 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 must be reported, along with patient demographic information as specified below to the Genetic Diseases Program Office by fax at (504) 219-4452. Receipt of faxed results must be verified by call to the Genetics Office at (504) 219-4413.

b. Described below are specific time deadlines after data reduction and interpretation for reporting positive results indicating probable disease to the Genetics Office. Laboratories must make arrangements with the Genetics Office for reporting after hours, weekends and holidays for positive results from tandem mass spectrometry and the assays for galactosemia, congenital adrenal hyperplasia and congenital hypothyroidism. Notification of presumptive positive results for biotinidase deficiency, sickle cell disease and cystic fibrosis will be made at the beginning of the next business day.

i. Metabolic disorders identified by tandem mass spectrometry and for galactosemia -

report results by 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.

vi. Cystic Fibrosis—report within 24 hours.

c. The specified information to be reported:

i. child's name
ii. parent or guardian's name
iii. child's street address
iv. child's date of birth
v. child's sex
vi. child's race
vii. parent's telephone number
viii. collection date
ix. test results
x. primary care physician
xi. age at collection (< or > 48 hours old)
xii. birth weight
xiii. full term or Premature
xiv. transfusion

Yes Date of last transfusion _____ No _____ (if available)

9. Provision of follow-up services. To ensure that reporting time deadlines are met for every positive result indicating probable disease under b above, 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 5 days, a home visit should be made;

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 must 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 Genetics Office address in Subsection G.8).

b. The laboratory must 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 will 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).
xv. child's test results

11. The laboratory must register by letter with the Genetic Diseases Program of the Office of Public Health each year. This letter must contain the following and be received in the Genetic Diseases Program Office by February 1 each year:

a. assurance of compliance with the requirements described in Subsection G1.-10 above.
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
e. reporting format for positive/abnormal test results

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

1. The laboratory reporting the results to the submitter must indicate that transfusion may alter all newborn screening results along with the following instruction:

Repeat Testing Recommended: 3 days after last transfusion
And 90 days after last transfusion

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

AUTHORITY NOTE: Promulgated in accordance with R.S. 40:1299, et seq.

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, 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: (May 2003), LR 32: 248 (February, 20, 2006), LR 34:13 (January 20, 2008), LR 34:442 (March 20, 2008)