**RULE**

**Louisiana Department of Health**

**Office of Public Health**

Under the authority of R.S. 40:4 and 40:5, and in accordance with R.S. 49:950 *et seq.*, the Administrative Procedure Act, notice is hereby given that the state health officer, acting through the Louisiana Department of Health, Office of Public Health (LDH/OPH), has recodified parts of Chapter 63 of Title 48—Public Health—General.

 This rule amends §6303 to include Spinal Muscular Atrophy (SMA), Mucopolysaccharidosis (MPS I), and Glycogen Storage Diseases Type II (Pompe Disease) to the Louisiana Newborn Screening Panel. The rule also amends the laboratory testing methodology used for newborn heel stick screening.

**TITLE 48**

**PUBLIC HEALTH - GENERAL**

Part V. Preventive Health Services

Subpart 18. Disability Prevention Program

Chapter 63. Newborn Heel Stick Screening

§6303. Purpose, Scope, and Laboratory Testing Methodology

A. 1.—8.a. …

 9. Neuromuscular Disorders:

 a. spinal muscular atrophy (SMA)

 10. Lysosomal Storage Disorders

 a. mucopolysaccharidosis type I (MPS I)

 b. glycogen storage disease type II (Pompe)

B. 1.—3. …

4. To ensure that specimens for testing are received within 24 to 48 hours or 1 to 2 days after collection, a state run courier will pick up specimens from each birthing facility and transport the specimens to the Office of Public Health (OPH) Laboratory. Specimens collected by other laboratories approved by OPH to perform newborn screening pursuant to the requirements of this Chapter, shall provide mailing envelopes to submitting hospitals which guarantee a delivery time no longer than ~~3~~2 days from mailing. ~~An~~ The use of the United States Postal Service and all other companies and courier services providing the required level of service stated herein are acceptable. C. …

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 screening specimen should be collected between 24 and 48 hours after birth.

 a. If the newborn is admitted or readmitted to the hospital within the first 28 days of life, the admitting facility shall collect and submit the newborn screening specimen unless proof of a previous normal newborn screening specimen result is available.

 b. If the newborn transferred from one facility to another, the transferring facility shall collect the newborn screening specimen and notify the next facility that the newborn screening specimen has been collected. The facility transporting a sick newborn should have the initial newborn screen documented in the newborn’s medical record. The receiving facility should determine if the newborn screen was done. If not, the newborn shall have an initial newborn screen collected upon admission.

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 2-5 days 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. 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.

4. Policy for Result Reporting and Repeat Screening Post Transfusion. Whenever possible, a specimen should be collected prior to transfusion. Repeat testing is recommended 3 days after transfusion and 90 days after last transfusion. 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.

 Please contact the Louisiana Genetic Diseases program for guidance on any other testing concerns.

5. 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.—D.1 …

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. If the laboratory determines the specimen to be unsatisfactory, the submitter shall collect and submit a second sample as soon as possible. If the newborn has been discharged, the submitter shall contact the newborn’s primary care provider or parent or guardian to collect a second sample. Training on collecting adequate specimens is available on the Newborn Screening website at ldh.la.gov/newborn.

F.—G.4. …

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** |
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| **Disease** | **Testing Methodology** |
| Disorders of Amino Acid Metabolism Disorders of Fatty Acid MetabolismDisorders 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 assayTotal Galactose |
| Hemoglobinopathies (Sickle Cell Diseases) | Cellulose acetate/citrate agarCapillary isoelectric focusing (CIEF)Gel isoelectric focusing (IEF)High Pressure Liquid Chromatography(HPLC)DNA Mutational AnalysisSickle Dex – is NOT AcceptableControls must include: F, A, S, C, D, EIf 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 phenotypePositive/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 |
| Cystic Fibrosis | Primary: Immunoreactive Trypsinogen; Time-Resolved fluoroimmunoassaySecond 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 TestQualitative Sweat Conductivity Test is NOT recommended. |
| Severe Combined Immunodeficiencies (SCID)Spinal Muscular Atrophy (SMA) | Real Time Quantitative Polymerase Chain Reaction (RTQPCR)  |
| Mucopolysaccharidosis type I (MPS I)Glycogen storage disease type II (Pompe) | Digital microfluidics |

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

P.O. Box 60630

New Orleans, Louisiana 70160-0630

5.b.—8.b. …

 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. 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 or gestational age; and

 xiv. transfusion given?

 Yes \_\_\_\_ No \_\_\_\_

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

 xv. Feeding type: human milk, formula (type), both (formula type)

9.—11.e. …

AUTHORITY NOTE: Promulgated in accordance with R.S. 40:1081.1 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 , 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), LR 48: