Notice of Intent

**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), intends to recodify parts of Chapter 63 of Title 48—Public Health—General.

 This proposed rule will amend §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 proposed rule will also amend 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 1 (MPS 1)

 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** |
| --- |
| **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:

**Family Impact Statement**

The proposed Rule is anticipated to have a known or foreseeable impact on family formation, stability, and autonomy. In particular, for newborns diagnosed with any of these conditions and their families, the proposed Rule may have a known or foreseeable impact on:

1. The stability of the family: the purpose of newborn screening is to identify genetic conditions which are treatable, life enhancing and potentially life-saving. Newborn screening is a very important service to families in detecting diseases at birth which can be identified through proper and available screening. Adding these additional diseases to the newborn screening panel would enhance the stability of the family by detecting this devastating condition early and preventing the negative health consequences.

2. The authority and rights of persons regarding the education and supervision of their children: This will not affect the authority, rights or supervision of parents over their children. Parents have the choice to “opt out” of the testing.

3. The functioning of the family: if detected and treated early, children affected by these disorders can lead long and fulfilling lives. This will contribute to a positive family structure.

4. Family earnings and family budget: testing for SMA, MPS I and Pompe Disease is life-saving as well as cost saving for families. Testing for these conditions will be added to the existing newborn screening blood spot panel at no cost to families, leading to early identification of the conditions. This testing saves lives and saves money. A late diagnosed case of SMA can result in up to $200,000 per year per case in medical bills and can ultimately lead to death within the first two years of life. Treatment for SMA with FDA approved methods is approximately $2,000,000 for a single-dose curative treatment and is covered by Medicaid and most commercial health insurance plans and results in a typical life for people with this condition. Without early testing and treatment, the life span of a person with MPS I is 10 years and Pompe Disease can cause death in the first year of life. Although there is no cure for these conditions, both can be managed with enzyme replacement therapy (ERT) which is covered by Louisiana Medicaid.

5. The behavior and personal responsibility of children: The addition of SMA, MPS I and Pompe would help children affected with these conditions lead typical lives.

6. The ability of the family or a local government to perform the function as contained in the proposed Rule: All children in Louisiana are tested at birth for most conditions recommended by the U.S. Department of Health and Human Services’ Advisory Committee on Heritable Disorders in Newborns and Children. Adding the new disorders to the newborn screening panel will not call for any additional effort of families or local governments.

**Poverty Impact Statement**

The proposed Rule is anticipated to have a known or foreseeable impact on any child, individual or family as defined by R.S. 49:973(B). In particular, for newborns diagnosed with any of these conditions and their families, there may be a foreseeable effect on:

1. The effect on household income, assets, and financial security: there will be a positive effect on household income, assets and financial security through the avoidance of health issues for families of children who have these additional diseases and were detected at birth. Health issues, if not treated early in life, can have huge financial impact to a family as they will have extreme medical costs.

2. The effect on early childhood development and preschool through postsecondary education development: if detected and treated early, children can develop without the burden of continued medical issues. When these diseases are not identified early, illness and eventually death, will likely occur before the child ever reaches pre-school.

3. The effect on employment and workforce development: there could be effect on the employment and workforce development of parents of children with these additional diseases. Caring for a very sick child could make employment for both parents difficult.

4. The effect on taxes and tax credits: there will be no effect on taxes and tax credits.

5. The effect on child and dependent care, housing, health care, nutrition, transportation, and utilities assistance: : there will be a positive effect on child and dependent care, housing, health care, nutrition, transportation, and utilities assistance. Early detection and treatment mean that a child, and their family, can operate without the burden of increased health care costs. Additionally, lost employment and strained resources for utilities can be avoided, as well as increased difficulties for child and dependent care, due to frequent illnesses.

**Small Business Analysis**

The proposed Rule should have no adverse impact on small businesses as defined in the Small Business Protection Act.

**Provider Impact Statement**

The proposed Rule should not have any known or foreseeable impact on providers as defined by HCR 170 of the 2014 Regular Legislative Session. Per HCR 170, "provider" means an organization that provides services for individuals with developmental disabilities. In particular, the impact is anticipated as follows:

1. there is no anticipated impact on the staffing level requirements or qualifications required to provide the same level of service as is currently being provided to the population of individuals with developmental/intellectual disabilities being served by the provider;

2. the total direct and indirect effect on the cost to the providers to provide the same level of service is anticipated to be minimal due to the low incidence rates for these conditions. The goal of testing newborns is to identify positive cases early in life and institute treatment as soon as possible; thus, more costly interventions later in life to babies who are not tested and are later found to exhibit symptoms of this disease are averted. The incidence for SMA is 1 per 10,000 births. Louisiana would expect to see 4-5 cases per year. The incidence for MPS I is 1 in 54,000 births and Louisiana would expect to see 1 case per year. The incidence of Pompe Disease is 1 in 28,000 births and Louisiana would expect to see about 2-3 cases per year; and

3. there is no anticipated impact on the overall effect on the ability of the provider to provide the same level of service.

**Public Comments**

Interested persons may submit written comments on the proposed rule. Such comments must be received no later than Tuesday, January 25, 2022 and should be addressed to Amy Zapata, MPH, Louisiana Department of Health, Office of Public Health - Bureau of Family Health, 1450 Poydras Street, Room 2013, New Orleans, LA 70112.

**Public Hearing**

Interested persons may submit a written request to conduct a public hearing by U.S. mail to the Office of the Secretary ATTN: LDH Rulemaking Coordinator, Post Office Box 629, Baton Rouge, LA 70821-0629; however, such request must be received no later than Monday, January 10, 2022**.** If the criteria set forth in R.S. 49:953(A)(2)(a) are satisfied, LDH will conduct a public hearing at 9:00AM on Tuesday, January 25, 2022 in Room 173 of the Bienville Building, which is located at 628 North Fourth Street, Baton Rouge, LA. To confirm whether or not a public hearing will be held, interested persons should first call Allen Enger at (225) 342-1342 after Monday, January 10, 2022. If a public hearing is to be held, all interested persons are invited to attend and present data, views, comments, or arguments, orally or in writing. In the event of a hearing, parking is available to the public in the Galvez Parking Garage which is located between North Sixth and North Fifth/North and Main Streets (cater-corner from the Bienville Building). Validated parking for the Galvez Garage may be available to public hearing attendees when the parking ticket is presented to the Bienville Building’s front security desk.

Dr. Courtney N. Phillips

Secretary

**FISCAL AND ECONOMIC IMPACT STATEMENT**

**FOR ADMINISTRATIVE RULES**

Person

Preparing

Statement: Cheryl L. Harris Dept.: Louisiana Department of Health

Phone: 504-568-8254 Office: Office of Public Health

Return

Address: \_1450 Poydras St\_Ste 2046

 New Orleans, LA 70112 Rule Title: Newborn Screening Rule (LAC 48.V.6303)\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

 Date Rule Takes Effect: Upon Promulgation

SUMMARY

In accordance with Section 953 of Title 49 of the Louisiana Revised Statutes, there is hereby submitted a fiscal and economic impact statement on the rule proposed for adoption, repeal or amendment. THE FOLLOWING STATEMENTS SUMMARIZE ATTACHED WORKSHEETS, I THROUGH IV AND WILL BE PUBLISHED IN THE LOUISIANA REGISTER WITH THE PROPOSED AGENCY RULE.

I. ESTIMATED IMPLEMENTATION COSTS (SAVINGS) TO STATE OR LOCAL GOVERNMENTAL UNITS

 There are no implementation costs or savings to local government.

The estimated costs to state government are expected to be $729,105 for FY22, $1,589,417 for FY23, and $1,759,788 for FY24. The cost of publication in the Louisiana Register is expected to be approximately $609.

II. ESTIMATED EFFECT ON REVENUE COLLECTIONS OF STATE OR LOCAL GOVERNMENTAL UNITS

There is no revenue impact on local governments.

Regarding state government, The Office of Public Health (OPH) is negotiating with the Louisiana Medicaid to reimburse OPH on the cost of the screening. At this time, the agency is unable to provide the exact dollar amount of the anticipated revenue increase. However, it is anticipated that the revenue will be equal or close to the expenditures.

III. ESTIMATED COSTS AND/OR ECONOMIC BENEFITS TO DIRECTLY AFFECTED PERSONS, SMALL BUSINESSES OR NON-GOVERNMENTAL GROUPS (Summary)

The benefit of this rule to affected persons is the reduction in the risk of mortality and cost of treatment associated with these conditions.

IV. ESTIMATED EFFECT ON COMPETITION AND EMPLOYMENT

The LDH Office of Public Health is the only entity in the state with the resources to provide this service. There is no estimated effect on competition or employment.

Signature of Agency Head or Designee Legislative Fiscal Officer or Designee

Kim Hood, JD

Assistant Secretary, Office of Public Health

Typed Name & Title of Agency Head or Designee

Date of Signature Date of Signature