

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

- 1) Heading of the Part: Newborn Metabolic Screening and Treatment Code
- 2) Code Citation: 77 Ill. Adm. Code 661
- 3)

<u>Section Numbers</u> :	<u>Adopted Action</u> :
661.10	Amended
661.15	Amended
661.30	Amended
661.35	Amended
661.40	Amended
661.50	Amended
661.70	Amended
- 4) Statutory Authority: Newborn Metabolic Screening Act [410 ILCS 240]
- 5) Effective Date of Rulemaking: January 19, 2012
- 6) Does this rulemaking contain an automatic repeal date? No
- 7) Do these amendments contain incorporations by reference? No
- 8) A copy of the adopted amendments, including any material incorporated by reference, is on file and available for public inspection at the Illinois Department of Public Health, 525 W. Jefferson Street, Springfield, Illinois 62761-0001.
- 9) Notice of Proposal Published in the Illinois Register: 35 Ill. Reg. 12668; July 29, 2011
- 10) Has JCAR issued a Statement of Objection to this rulemaking? No
- 11) Differences between proposal and final version: Two nonsubstantive grammatical changes were made.
- 12) Have all the changes agreed upon by the agency and JCAR been made as indicated in the agreements issued by JCAR? No agreements were necessary.
- 13) Will this rulemaking replace any emergency rulemaking currently in effect? No
- 14) Are there any amendments pending on this Part? No

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

15) Summary and Purpose of Amendments: These amendments include provisions to describe the mandate for testing all infants born in Illinois for severe combined immunodeficiency (SCID). Newborn screening is described by the Newborn Metabolic Screening Act, enacted in 2007 (Public Act 95-695). The adopted rulemaking defines qualifications for the physician specialists who will be providing follow-up care for children identified through newborn screening with a possible diagnosis of SCID. General recommended treatments for infants affected with SCID are also defined, and the current newborn screening fee has been increased to cover the cost of testing for SCID.

16) Information and questions regarding these adopted amendments shall be directed to:

Susan Meister
Division of Legal Services
Illinois Department of Public Health
535 West Jefferson, Fifth Floor
Springfield, Illinois 62761

217/782-2043
(E-mail: dph.rules@illinois.gov)

The full text of the Adopted Amendments begins on the next page:

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

TITLE 77: PUBLIC HEALTH
CHAPTER I: DEPARTMENT OF PUBLIC HEALTH
SUBCHAPTER i: MATERNAL AND CHILD HEALTHPART 661
NEWBORN METABOLIC SCREENING AND TREATMENT CODE

Section

661.10	Responsibility
661.15	Definitions
661.20	Collection of Blood and Submission of Specimens
661.30	Interpretation of Results
661.35	Designation of Medical Specialists
661.40	Reports
661.50	Diagnosis and Treatment
661.60	Exemption
661.70	Fee Assessment and Payment

AUTHORITY: Implementing and authorized by the Newborn Metabolic Screening Act [410 ILCS 240].

SOURCE: Adopted December 14, 1973; emergency rules at 3 Ill. Reg. 28, p. 224, effective June 28, 1979, for a maximum of 150 days; rules repealed and new rules adopted at 3 Ill. Reg. 48, p. 42, effective November 20, 1979; amended at 5 Ill. Reg. 4593, effective April 15, 1981; amended and codified at 8 Ill. Reg. 19041, effective September 26, 1984; amended at 11 Ill. Reg. 12921, effective August 1, 1987; amended at 13 Ill. Reg. 15079, effective October 1, 1989; amended at 14 Ill. Reg. 13292, effective August 15, 1990; amended at 17 Ill. Reg. 13609, effective August 1, 1993; amended at 19 Ill. Reg. 15720, effective November 1, 1995; expedited correction at 20 Ill. Reg. 3590, effective November 1, 1995; amended at 22 Ill. Reg. 20639, effective November 10, 1998; amended at 26 Ill. Reg. 10676, effective July 1, 2002; amended at 26 Ill. Reg. 18412, effective January 1, 2003; amended at 31 Ill. Reg. 13203, effective August 28, 2007; amended at 34 Ill. Reg. 940, effective December 31, 2009; amended at 36 Ill. Reg. 1753, effective January 19, 2012.

Section 661.10 Responsibility

- a) The physician in attendance at or immediately after the birth of the newborn infant shall have primary responsibility for seeing that a specimen of the infant's blood is screened in accordance with this Part. Newborn screening includes tests

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

for the following disorders: classical phenylketonuria (PKU) and certain other amino acid, organic acid, and fatty acid oxidation disorders; primary hypothyroidism; classical galactosemia; congenital adrenal hyperplasia due to 21-hydroxylase deficiency; biotinidase deficiency; sickle cell disease/trait; cystic fibrosis; ~~and~~ lysosomal storage disorders; ~~and severe combined immunodeficiency~~. Specific diseases in the categories of amino acid, organic acid, and fatty acid oxidation ~~disorders~~ and lysosomal storage disorders shall be reviewed by the Genetic and Metabolic Diseases Advisory Committee. The Department ~~will~~ consider the recommendations of the Genetic and Metabolic Diseases Advisory Committee in determining to include an additional disorder in the screening panel. Implementation of the Department's determination ~~is~~ subject to that determination's adoption by rule. For a current list of disorders, refer to the Illinois Department of Public Health Newborn Screening Practitioner's Manual. A blood specimen meeting the requirements for testing shall suffice for all tests (see Section 661.20). The physician may delegate this responsibility to the hospital administrator or to the administrator's designated representative, such as a member of the pediatrics staff, the laboratory director, the obstetrical supervisor, or other hospital official.

- b) If the infant is not born in or admitted to a hospital or when there is no physician in attendance at or immediately after the birth, the physician caring for the infant during the first month of life shall be the individual responsible for seeing that a blood specimen for newborn screening is submitted. When there is no physician caring for ~~the~~ such an infant during this period, the parents or guardian is responsible. Local health authorities or the Department ~~will~~ assist the parents or guardian in having a blood specimen submitted for testing.
- c) All specimens collected pursuant to this Part shall be submitted for testing to the Newborn Screening Section, Division of Laboratories, Illinois Department of Public Health, 2121 West Taylor Street, Chicago, Illinois 60612 (see Section 661.20).
- d) When a retest is determined to be necessary pursuant to Section 661.30 of this Part, the Illinois Department of Public Health ~~will~~ notify the physician or his or her designee who is responsible for obtaining another specimen and having the specimen tested.

(Source: Amended at 36 Ill. Reg. 1753, effective January 19, 2012)

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

Section 661.15 Definitions

"Act" means the Newborn Metabolic Screening Act [410 ILCS 240].

"Advisory Committee" means the Genetic and Metabolic Diseases Advisory Committee appointed by the Director.

"CF" means cystic fibrosis.

"CLSI" means Clinical and Laboratory Standards Institute.

"Department" or "DPH" means the Department of Public Health.

"Director" means the Director of the Department of Public Health.

"Formula" means a medically prescribed treatment substance that has been designed to treat a specific metabolic disorder.

"LSD" means lysosomal storage disorders, including the following: Krabbe, Pompe, Gaucher, Fabry, and Niemann-Pick, which are inherited metabolic disorders caused by lysosomal dysfunction, usually as a consequence of deficiency of a single enzyme required for the metabolism of lipids, glycoproteins or mucopolysaccharides.

"Newborn screening" or "testing" means the testing of a blood sample for classical phenylketonuria (PKU) and certain other amino acid, organic acid, and fatty acid oxidation disorders, primary hypothyroidism, classical galactosemia, congenital adrenal hyperplasia due to 21-hydroxylase deficiency, biotinidase deficiency, sickle cell disease/trait, cystic fibrosis, and lysosomal storage disorders, and severe combined immunodeficiency. At times, variant forms of some disorders, or related conditions, may also be identified.

"PKU" means classical phenylketonuria.

"Tandem mass spectrometry" means use of a tandem mass spectrometer and associated software to test a newborn screening sample.

"MS/MS" means Tandem Mass Spectrometry.

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

"SCID" means severe combined immunodeficiency and T cell lymphopenia.

"Using accepted statistical techniques" means using techniques that have been published in peer reviewed scientific literature.

(Source: Amended at 36 Ill. Reg. 1753, effective January 19, 2012)

Section 661.30 Interpretation of Results

Although the majority of infants affected by disorders included in the newborn screening panel will be identified by this screening, due to genetic variabilities and variations in health status, specimen quality, and timing of specimen collection, not all infants affected by ~~such~~ a disorder may be identified. As with any laboratory test, false positive and false negative results are possible. Newborn screening test results are insufficient information on which to base diagnosis or treatment. Tests will be conducted at a Department of Public Health laboratory designated to perform the tests (Section 2(e) of the Act), as follows:

- a) Phenylketonuria
 - 1) Normal phenylalanine levels shall be established using accepted statistical techniques.
 - 2) When the blood phenylalanine level is deemed to be abnormal, the Department ~~will~~shall recommend a repeat newborn screening test or referral of the infant to a designated medical specialist for a quantitative phenylalanine determination and other diagnostic studies as determined by the medical specialist.
- b) Primary Hypothyroidism
 - 1) Neonatal levels for thyroid stimulating hormone (TSH) vary with gestational age, ~~birth weight~~birthweight, time of collection and in response to concurrent medical problems. Normal TSH and normal thyroxine (T4) levels shall be established using accepted statistical techniques.
 - 2) When the TSH level or the T4 level is deemed to be abnormal, the Department ~~will~~shall recommend a repeat newborn screening test or referral of the infant to a designated pediatric endocrinologist for further evaluation for primary hypothyroidism and additional serum testing for

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

thyroid function.

c) Galactosemia

- 1) Laboratory tests for galactosemia may be performed by testing for total galactose (galactose and galactose-1-phosphate) or a deficiency of the galactose-1-phosphate uridyl transferase enzyme. Normal test results indicate a normal level of total galactose or the presence of the enzyme. Test results are abnormal when the level of total galactose is above the normal range or the presence of the enzyme is not detected. Normal ranges shall be established using accepted statistical techniques.
- 2) When the galactose or enzyme levels are deemed abnormal, recommendations may be given to change the diet of the infant to a galactose free diet. The Department ~~will~~shall recommend a repeat newborn screening test or referral of the infant to a designated medical specialist for further diagnostic studies.

d) Congenital Adrenal Hyperplasia (secondary to 21-hydroxylase deficiency)

- 1) Neonatal levels for 17-hydroxyprogesterone vary with gestational age, ~~birth weight~~birthweight, time of collection and in response to concurrent medical problems. Normal 17-hydroxyprogesterone levels shall be established using accepted statistical techniques.
- 2) When the 17-hydroxyprogesterone level is deemed to be abnormal, the Department ~~will~~shall recommend a repeat newborn screening test or referral of the infant to a designated pediatric endocrinologist for further evaluation for congenital adrenal hyperplasia.

e) Biotinidase Deficiency

- 1) Laboratory tests for biotinidase deficiency are designed to detect a deficiency of the biotinidase enzyme. Normal test results indicate the presence of the enzyme. Test results are abnormal when the presence of the enzyme is not detected.
- 2) When the determination of the enzyme is deemed abnormal, the Department ~~will~~shall recommend a repeat newborn screening test or

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

referral of the infant to a designated medical specialist for a quantitative determination of the biotinidase enzyme and further diagnostic studies.

- f) Sickle Cell Disease/Trait and Other Hemoglobinopathies
Qualitative testing will determine the presence of A, F, S, C and other hemoglobins.
- 1) When F and S hemoglobins, but no A hemoglobin, are detected on the same specimen, the Department ~~will~~shall recommend referral to a designated medical specialist for follow-up and genetic counseling.
 - 2) When F, S and C hemoglobins, but no A hemoglobin, are detected on the same specimen, the Department ~~will~~shall recommend referral to a designated medical specialist for follow-up and genetic counseling.
 - 3) When F, A and C hemoglobins or F, A and S hemoglobins are detected on the same specimen, the Department ~~will~~shall recommend parental testing and genetic counseling by the attending physician or another qualified counselor.
 - 4) When A hemoglobin is detected as the predominant hemoglobin, and the specimen was collected at less than ~~two~~2 months of age, ~~it will be assumed that~~ the infant ~~will be assumed to have~~ received a blood transfusion, and a report indicating ~~that the infant received a blood transfusion~~such will be made. A repeat newborn screening specimen should be drawn from all such infants ~~three~~3 months post-transfusion.
- g) Phenylketonuria (PKU) and ~~Other Amino Acid~~other amino acid, ~~Organic Acid~~organic acid, and ~~Fatty Acid Oxidation Disorders~~fatty acid oxidation disorders (~~Note~~-PKU testing is described in Section 661.30(a).):
- 1) Analysis shall be performed by MS/MS. The patient metabolite distribution patterns shall be compared to normal populations. Pattern analysis, and internal metabolite ratios relative to normal populations, shall be calculated using accepted statistical techniques.
 - 2) When blood levels or ratios are found to be abnormal, indicating the possibility of a metabolic condition harmful to the infant, the Department ~~will~~shall recommend a repeat newborn screening test or referral of the

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

infant to a designated medical specialist for appropriate definitive testing and diagnostic studies.

- h) Cystic Fibrosis (CF)
- 1) CF is indicated by elevated neonatal levels of immunoreactive trypsinogen (IRT) that can be detected in dried blood spots by immunoassay or other techniques. The normal IRT range shall be established using accepted statistical techniques.
 - 2) When elevated levels of IRT are detected, testing by genetic mutation analysis shall be performed in order to decrease false positive results. Because there are over 1,000 mutations in the CF transmembrane conductance regulator (CFTR) gene, testing will yield only 90 to 95 percent sensitivity.
 - 3) When IRT levels and/or mutation analysis are found to be abnormal, thus indicating the possibility of CF, the Department ~~will~~ recommend referral of the infant to a designated medical specialist for appropriate definitive testing and diagnostic studies.
- i) Lysosomal Storage Disorders (LSDs)
- 1) An LSD can be detected in dried blood spots by using tandem mass spectrometry or other methods. Normal testing parameters shall be established using accepted statistical techniques.
 - 2) When testing parameters are found to be abnormal, thus indicating the possibility of an LSD, the Department will recommend referral of the infant to a designated medical specialist for appropriate definitive testing and diagnostic studies.
 - 3) ~~After an initial phase-in project to establish normal testing parameters and validate the screening, all specimens~~ After an initial phase-in project to establish normal testing parameters and validate the screening, all specimens ~~To establish normal testing parameters and validate the screening technique, a phase-in project will be conducted from November 1, 2010 through May 31, 2011 requiring LSD screening of all babies born at the University of Chicago Hospitals and Northwestern Memorial Hospital. At the conclusion of the phase-in project, all specimens~~ submitted to the Illinois Department of Public

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

Health Newborn Screening Laboratory will be tested for LSDs.

- j) Severe Combined Immunodeficiency (SCID)
- 1) SCID can be detected in dried blood spots by using DNA-based methods, such as polymerase chain reaction (PCR) or other methods. Normal testing parameters shall be established using accepted statistical techniques.
 - 2) When testing parameters are found to be abnormal, thus indicating the possibility of SCID, the Department will recommend referral of the infant to a designated medical specialist for appropriate definitive testing and diagnostic studies
 - 3) To establish normal testing parameters and validate the screening technique, a phase-in project will be conducted for a six-month period after January 1, 2012, and will require SCID screening of all babies born at a small group of birthing hospitals to be designated. At the conclusion of the phase-in project, all specimens submitted to the Illinois Department of Public Health Newborn Screening Laboratory will be tested for SCID.

(Source: Amended at 36 Ill. Reg. 1753, effective January 19, 2012)

Section 661.35 Designation of Medical Specialists

- a) The Newborn Screening Program Manager, with the advice of the Director of the University of Illinois Division of Specialized Care for Children, and the Chairman of the Advisory Committee, ~~will~~ shall designate qualified professionals to serve as medical specialists in specified disease categories within the Newborn Screening Program. These medical specialists should provide care to children identified through newborn screening in collaboration with the primary care provider.
- b) Equivalency in all qualifications specified in this Section ~~will~~ shall be determined by the Newborn Screening Program Manager, with the advice of the Director of the University of Illinois Division of Specialized Care for Children, and the Chairman of the Advisory Committee.
- c) The minimum qualifications required for designation as a medical specialist are a license to practice medicine in all its branches in Illinois, or licensure in the state

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

of practice, and certification by the American Board of Pediatrics or equivalent board from another country. In addition, to be designated to serve in specified disease categories, medical specialists shall also have the following qualifications:

- 1) Phenylketonuria (PKU) and All Other Disorders~~all other disorders~~ of Amino Acid~~amino acid~~ and Organic Acid Metabolism~~organic acid metabolism~~: certification by the American Board of Medical Genetics in Clinical Biochemical Genetics or certification by the American Board of Medical Genetics in Clinical Genetics with at least one year of experience post-training in the diagnosis and treatment of amino acid and organic acid disorders. The medical specialist shall have the capacity to provide a multidisciplinary approach to care, including the availability on site~~on-site~~ of specially trained metabolic dieticians and a biochemical genetics laboratory; for citrullinemia and argininosuccinic aciduria, medical specialists should have on-site availability of required medical therapies, such as hemodialysis, that are necessary for the treatment of patients with these disorders.
- 2) Primary Hypothyroidism: training in Pediatric Endocrinology with membership in the Lawson Wilkins Pediatric Endocrinology Society or certification of special competence in Pediatric Endocrinology by the American Board of Pediatrics.
- 3) Galactosemia: certification by the American Board of Medical Genetics in Clinical Biochemical Genetics or certification by the American Board of Medical Genetics in Clinical Genetics with at least one year of experience post-training in the diagnosis and treatment of galactosemia and inborn errors of metabolism. Medical specialists should have the capacity to provide a multidisciplinary approach to care, including the availability on site~~on-site~~ of specially trained metabolic dieticians.
- 4) Congenital Adrenal Hyperplasia: training in Pediatric Endocrinology with membership in the Lawson Wilkins Pediatric Endocrinology Society or certification of special competence in Pediatric Endocrinology by the American Board of Pediatrics.
- 5) Biotinidase Deficiency: certification by the American Board of Medical Genetics in Clinical Biochemical Genetics or certification by the American Board of Medical Genetics in Clinical Genetics with at least one

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

year of experience post-training in the diagnosis and treatment of biotinidase deficiency and inborn errors of metabolism. Medical specialists should have the capacity to provide a multidisciplinary approach to care, including the availability on site~~on-site~~ of specially trained metabolic dieticians.

- 6) Sickle Cell Disease: training in Pediatric Hematology and certification of special competence in Pediatric Hematology-Oncology by the American Board of Pediatrics.
- 7) Fatty Acid Oxidation Disorders: certification by the American Board of Medical Genetics in Clinical Biochemical Genetics or certification by the American Board of Medical Genetics in Clinical Genetics with at least one year of experience post-training in the diagnosis and treatment of fatty acid oxidation disorders. Medical specialists should have the capacity to provide a multidisciplinary approach to care, including the availability on site~~on-site~~ of specially trained metabolic dieticians.
- 8) Cystic Fibrosis: certification by the American Board of Pediatrics in Pediatric Pulmonology or Pediatric Gastroenterology. Medical specialists should provide prompt access to quantitative pilocarpine iontophoresis sweat chloride testing in a laboratory that meets all CLSI standards. Medical specialists should provide a multidisciplinary approach to care, including the availability of on-site genetic counselors, dieticians, respiratory therapists and social workers. Medical specialists should provide access to microbiology laboratories that use CF-specific protocols for detection of respiratory tract infection.
- 9) Lysosomal Storage Disorders: certification by the American Board of Medical Genetics in Clinical Biochemical Genetics or certification by the American Board of Medical Genetics in Clinical Genetics with at least one year of experience post-training in the diagnosis and treatment of LSDs. Medical specialists should have the capacity to provide enzyme replacement infusion therapies and to provide a multidisciplinary approach to care, including the availability of pediatric specialists in neurology, cardiology and pulmonology. In addition to the above requirements, for Krabbe disease, medical specialists should be affiliated with a facility that has experience in performing stem cell transplantation.

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

- 10) Severe Combined Immunodeficiency and T Cell Lymphopenia: certification by the American Board of Allergy and Immunology with at least one year post-training in the diagnosis and treatment of primary immunodeficiency diseases. Medical specialists should have the capacity to diagnose SCID, DiGeorge syndrome or other causes of T cell lymphopenia and to provide a multidisciplinary approach to treatment, including access to specialists in stem cell transplantation, and be affiliated with a facility that has experience in performing stem cell transplantation.

(Source: Amended at 36 Ill. Reg. 1753, effective January 19, 2012)

Section 661.40 Reports

- a) Only collection forms with attached filter paper blood collectors supplied by the Division of Laboratories, Illinois Department of Public Health, ~~2121 West Taylor Street, Chicago, Illinois 60612~~ are to be used in submitting blood specimens for newborn screening.
- b) Any hospital performing the required newborn screening tests in addition to submitting specimens to the Illinois Department of Public Health Laboratory shall comply with all requirements of this Part, and shall notify the Department immediately by telephone whenever testing on an infant indicates that:
- 1) phenylalanine levels are abnormal;
 - 2) T4 determinations are abnormal or TSH determinations are abnormal;
 - 3) total galactose or galactose-1-phosphate uridyl transferase determinations are abnormal;
 - 4) 17-hydroxyprogesterone determinations are abnormal;
 - 5) biotinidase enzyme determinations are abnormal;
 - 6) abnormal hemoglobin patterns are detected;
 - 7) abnormal amino acid or acylcarnitine patterns have been identified;

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

- 8) abnormal determinations that may indicate cystic fibrosis have been identified;
- 9) abnormal determinations that may indicate a lysosomal storage disorder have been identified;
- 10) abnormal determinations that may indicate severe combined immunodeficiency or T cell lymphopenia have been identified.

(Source: Amended at 36 Ill. Reg. 1753, effective January 19, 2012)

Section 661.50 Diagnosis and Treatment

The Department ~~will~~shall also maintain a registry to record the results of diagnosis and treatment for all diagnosed cases identified. ~~Ongoing~~It is imperative to perform ongoing evaluation of the newborn screening program is imperative. This process includes outcome evaluation of children diagnosed through newborn screening. The Department ~~will annually~~shall request updated information; from the medical specialist or primary care provider; ~~updated information annually~~; concerning developmental milestones; for each child diagnosed with a disorder for which the Department screens. The Department ~~will at all times~~shall maintain confidentiality at all times with regard to patient information.

- a) Phenylketonuria and Hyperphenylalaninemia. The Department will supply the necessary medically prescribed treatment formulas ~~will be supplied by the~~ Department for diagnosed cases as long as medically indicated. Long-term follow-up of children with phenylketonuria or hyperphenylalaninemia is necessary to adjust diet and to assess growth and development. Medical management by a designated medical specialist is required in order for a patient to receive treatment formulas from DPH. The administration of treatment formulas shall not be instituted until a complete amino acid analysis to corroborate the positive screening test has been performed, under the direction of a designated medical specialist, to establish the diagnosis of phenylketonuria.
- b) Primary Hypothyroidism. Medical management by a designated pediatric endocrinologist is highly recommended. Replacement therapy with thyroid hormone is currently the standard treatment. Long-term follow-up of children with primary hypothyroidism is necessary in order to adjust medication and to assess growth and development.

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

- c) Galactosemia. Medical management by a designated medical specialist is highly recommended. Therapy with a galactose free diet is currently the standard treatment. Long-term follow-up of children with galactosemia is necessary in order to ensure proper growth and development.
- d) Congenital Adrenal Hyperplasia. Medical management by a designated pediatric endocrinologist is highly recommended. Replacement therapy with glucocorticoids and, in some cases, mineralocorticoids is currently the standard treatment. Long-term follow-up of children with congenital adrenal hyperplasia is necessary in order to adjust medications and to assess growth and development.
- e) Biotinidase Deficiency. Medical management by a designated medical specialist is highly recommended. Therapy with pharmacological doses of biotin is required. Long-term follow-up of children with biotinidase deficiency is necessary in order to ensure proper growth and development.
- f) Sickle Cell Disease. Medical management by a designated pediatric hematologist-oncologist is highly recommended. Antibiotic prophylaxis and immunization to prevent pneumococcal infections are currently the standard treatment after a designated medical specialist has made a definitive diagnosis ~~has been made~~ of a sickling disease ~~by a designated medical specialist~~. Long-term follow-up of children with sickle cell disease is necessary in order to assess growth and development.
- g) Other Amino Acid, Organic Acid and Fatty Acid Oxidation Disorders. The Department will supply the necessary medically prescribed treatment formulas ~~will be supplied by the Department~~ for diagnosed cases as long as medically indicated. Long-term follow-up of children with these metabolic disorders is necessary to adjust diet and to assess growth and development. Medical management by a designated medical specialist is required in order for a patient to receive treatment formulas from DPH. Many of these disorders can be properly and supportively managed by dietary therapy. Ongoing care of these children will require long-term follow-up by the medical specialist to ensure proper development.
- h) Cystic Fibrosis. Medical management by a designated medical specialist is highly recommended. Prompt evaluation of exocrine pancreatic status coupled with nutritional counseling is recommended after diagnostic confirmation. Close

DEPARTMENT OF PUBLIC HEALTH

NOTICE OF ADOPTED AMENDMENTS

follow-up by a medical specialist is recommended to monitor and treat changes in nutrition and respiratory infection status.

- i) Lysosomal ~~Storage Disorders~~~~storage disorders~~. Medical management by a designated medical specialist is highly recommended. Enzyme replacement therapy or stem cell transplant have demonstrated benefits for patients with these disorders. Long-term follow-up of children with lysosomal storage disorders is necessary to monitor treatment and to assess growth and development.

- j) Severe Combined Immunodeficiency (SCID) and T Cell Lymphopenia. Medical management by a designated medical specialist is highly recommended to confirm the diagnosis of SCID or other cause of T cell lymphopenia and to start therapy as soon as possible. Adenosine deaminase-deficient SCID can be treated by enzyme replacement and immunoglobulin replacement therapies. All forms of SCID can be treated by stem cell transplantation, while a few forms of SCID can be treated by gene therapy. Complete DiGeorge syndrome can be treated by thymic transplantation. Long-term follow-up is necessary to document immune reconstitution and to assess growth and development.

(Source: Amended at 36 Ill. Reg. 1753, effective January 19, 2012)

Section 661.70 Fee Assessment and Payment

- a) Each institution or person submitting to the Department any sample for newborn screening shall be assessed a fee of ~~\$8878~~. When the Director makes a determination to add screening for any additional disorders in the LSD category, pursuant to Section 661.10, this fee shall be increased by \$2 for each disorder added.

- b) Statements of fee assessment shall be mailed on a monthly basis to facilities submitting specimens for analysis.

- c) Payment shall be rendered to the Department upon receipt of the monthly statement of fee assessment.

(Source: Amended at 36 Ill. Reg. 1753, effective January 19, 2012)