

1 AN ACT concerning health facilities.

2 **Be it enacted by the People of the State of Illinois,**
3 **represented in the General Assembly:**

4 Section 5. The Newborn Metabolic Screening Act is amended
5 by changing Sections 1, 1.5, and 2 and by adding Sections 1.10,
6 3.1, 3.2, and 3.3 as follows:

7 (410 ILCS 240/1) (from Ch. 111 1/2, par. 4903)

8 Sec. 1. The Illinois Department of Public Health shall
9 promulgate and enforce rules and regulations requiring that
10 every newborn be subjected to tests for genetic,
11 ~~phenylketonuria, hypothyroidism, galactosemia and such other~~
12 metabolic, and congenital anomalies ~~diseases~~ as the Department
13 may deem necessary ~~from time to time~~. The Department is
14 empowered to promulgate such additional rules and regulations
15 as are found necessary for the administration of this Act,
16 including mandatory reporting of the results of all tests for
17 these conditions to the Illinois Department of Public Health.

18 (Source: P.A. 83-87.)

19 (410 ILCS 240/1.5)

20 Sec. 1.5. Definitions. In this Act:

21 "Accredited laboratory" means any laboratory that holds a
22 valid certificate issued under the Clinical Laboratory

1 Improvement Amendments of 1988, 102 Stat. 2903, 42 U.S.C. 263a,
2 as amended, and that reports its screening results by using
3 normal pediatric reference ranges.

4 "Department" means the Department of Public Health.

5 ~~"Expanded screening" means screening for genetic and
6 metabolic disorders, including but not limited to amino acid
7 disorders, organic acid disorders, fatty acid oxidation
8 disorders, and other abnormal profiles, in newborn infants that
9 can be detected through the use of a tandem mass spectrometer.~~

10 ~~"Tandem mass spectrometer" means an analytical instrument
11 used to detect numerous genetic and metabolic disorders at one
12 time.~~

13 (Source: P.A. 92-701, eff. 7-19-02.)

14 (410 ILCS 240/1.10 new)

15 Sec. 1.10. Critical congenital heart disease.

16 (a) The General Assembly finds as follows:

17 (1) According to the United States Secretary of Health
18 and Human Services Advisory Committee on Heritable
19 Disorders in Newborns and Children, congenital heart
20 disease affects approximately 7 to 9 of every 1,000 live
21 births in the United States and Europe. The federal Centers
22 for Disease Control and Prevention state that critical
23 congenital heart disease is the leading cause of infant
24 death due to birth defects.

25 (2) Many newborn lives could potentially be saved by

1 earlier detection and treatment of critical congenital
2 heart disease if health care facilities in the State were
3 required to perform a simple, non-invasive newborn
4 screening in conjunction with current screening methods.

5 (b) The Department shall require that screening tests for
6 critical congenital heart defects be performed at birthing
7 hospitals and birth centers in accordance with a testing
8 protocol adopted by the Department, by rule, in line with
9 current standards of care, such as pulse oximetry screening,
10 and may authorize screening tests for additional congenital
11 anomalies to be performed at birthing hospitals and birth
12 centers in accordance with a testing protocol adopted by the
13 Department, by rule.

14 (c) The Department may authorize health care facilities to
15 report screening test results and follow-up information.

16 (410 ILCS 240/2) (from Ch. 111 1/2, par. 4904)

17 Sec. 2. General provisions. The Department of Public Health
18 shall administer the provisions of this Act and shall:

19 (a) Institute and carry on an intensive educational program
20 among physicians, hospitals, public health nurses and the
21 public concerning disorders included in newborn screening ~~the~~
22 ~~diseases phenylketonuria, hypothyroidism, galactosemia and~~
23 ~~other metabolic diseases.~~ This educational program shall
24 include information about the nature of the diseases and
25 examinations for the detection of the diseases in early infancy

1 in order that measures may be taken to prevent the ~~intellectual~~
2 disabilities resulting from the diseases.

3 (a-5) Require that ~~Beginning July 1, 2002, provide~~ all
4 newborns be screened ~~with expanded screening tests~~ for the
5 presence of certain genetic, metabolic, and congenital
6 anomalies as determined by the Department, by rule.

7 (a-5.1) Require that all blood and biological specimens
8 collected pursuant to this Act or the rules adopted under this
9 Act be submitted for testing to the nearest Department
10 laboratory designated to perform such tests. The following
11 provisions shall apply concerning testing:

12 (1) The Department may develop a reasonable fee
13 structure and may levy fees according to such structure to
14 cover the cost of providing this testing service and for
15 the follow-up of infants with an abnormal screening test.
16 Fees collected from the provision of this testing service
17 shall be placed in the Metabolic Screening and Treatment
18 Fund. Other State and federal funds for expenses related to
19 metabolic screening, follow-up, and treatment programs may
20 also be placed in the Fund.

21 (2) Moneys shall be appropriated from the Fund to the
22 Department solely for the purposes of providing newborn
23 screening, follow-up, and treatment programs. Nothing in
24 this Act shall be construed to prohibit any licensed
25 medical facility from collecting additional specimens for
26 testing for metabolic or neonatal diseases or any other

1 diseases or conditions, as it deems fit. Any person
2 violating the provisions of this subsection (a-5.1) is
3 guilty of a petty offense. ~~endocrine, or other metabolic~~
4 ~~disorders, including phenylketonuria, galactosemia,~~
5 ~~hypothyroidism, congenital adrenal hyperplasia,~~
6 ~~biotinidase deficiency, and sickling disorders, as well as~~
7 ~~other amino acid disorders, organic acid disorders, fatty~~
8 ~~acid oxidation disorders, and other abnormalities~~
9 ~~detectable through the use of a tandem mass spectrometer.~~

10 (3) If by July 1, 2002, the Department is unable to
11 provide the ~~expanded~~ screening using the State Laboratory,
12 it shall temporarily provide such screening through an
13 accredited laboratory selected by the Department until the
14 Department has the capacity to provide screening through
15 the State Laboratory. If ~~expanded~~ screening is provided on
16 a temporary basis through an accredited laboratory, the
17 Department shall substitute the fee charged by the
18 accredited laboratory, plus a 5% surcharge for
19 documentation and handling, for the fee authorized in this
20 subsection (a-5.1) ~~(e) of this Section.~~

21 (a-5.2) Maintain a registry of cases, including
22 information of importance for the purpose of follow-up services
23 to assess long-term outcomes.

24 (a-5.3) Supply the necessary metabolic treatment formulas
25 where practicable for diagnosed cases of amino acid metabolism
26 disorders, including phenylketonuria, organic acid disorders,

1 and fatty acid oxidation disorders for as long as medically
2 indicated, when the product is not available through other
3 State agencies.

4 (a-5.4) Arrange for or provide public health nursing,
5 nutrition, and social services and clinical consultation as
6 indicated.

7 (a-5.5) The Department shall utilize the Genetic and
8 Metabolic Diseases Advisory Committee established under the
9 Genetic and Metabolic Diseases Advisory Committee Act to
10 provide guidance and recommendations to the Department's
11 newborn screening program. The Genetic and Metabolic Diseases
12 Advisory Committee shall review the feasibility and
13 advisability of including additional metabolic, genetic, and
14 congenital disorders in the newborn screening panel, according
15 to a review protocol applied to each suggested addition to the
16 screening panel. The Department shall consider the
17 recommendations of the Genetic and Metabolic Diseases Advisory
18 Committee in determining whether to include an additional
19 disorder in the screening panel prior to proposing an
20 administrative rule concerning inclusion of an additional
21 disorder in the newborn screening panel. Notwithstanding any
22 other provision of law, no new screening may begin prior to the
23 occurrence of all the following:

24 (1) the establishment and verification of relevant and
25 appropriate performance specifications as defined under
26 the federal Clinical Laboratory Improvement Amendments and

1 regulations thereunder for U.S. Food and Drug
2 Administration-cleared or in-house developed methods,
3 performed under an institutional review board-approved
4 protocol, if required;

5 (2) the availability of quality assurance testing
6 methodology for the processes set forth in item (1) of this
7 subsection (a-5.5);

8 (3) the acquisition and installment by the Department
9 of the equipment necessary to implement the screening
10 tests;

11 (4) the establishment of precise threshold values
12 ensuring defined disorder identification for each
13 screening test;

14 (5) the authentication of pilot testing achieving each
15 milestone described in items (1) through (4) of this
16 subsection (a-5.5) for each disorder screening test; and

17 (6) the authentication of achieving the potential of
18 high throughput standards for statewide volume of each
19 disorder screening test concomitant with each milestone
20 described in items (1) through (4) of this subsection
21 (a-5.5).

22 (a-6) (Blank). ~~In accordance with the timetable specified~~
23 ~~in this subsection, provide all newborns with expanded~~
24 ~~screening tests for the presence of certain Lysosomal Storage~~
25 ~~Disorders known as Krabbe, Pompe, Gaucher, Fabry, and~~
26 ~~Niemann Pick. The testing shall begin within 6 months following~~

1 ~~the occurrence of all of the following:~~

2 ~~(i) the establishment and verification of relevant and~~
3 ~~appropriate performance specifications as defined under~~
4 ~~the federal Clinical Laboratory Improvement Amendments and~~
5 ~~regulations thereunder for Federal Drug~~
6 ~~Administration cleared or in house developed methods,~~
7 ~~performed under an institutional review board approved~~
8 ~~protocol, if required;~~

9 ~~(ii) the availability of quality assurance testing~~
10 ~~methodology for these processes;~~

11 ~~(iii) the acquisition and installment by the~~
12 ~~Department of the equipment necessary to implement the~~
13 ~~expanded screening tests;~~

14 ~~(iv) establishment of precise threshold values~~
15 ~~ensuring defined disorder identification for each~~
16 ~~screening test;~~

17 ~~(v) authentication of pilot testing achieving each~~
18 ~~milestone described in items (i) through (iv) of this~~
19 ~~subsection (a 6) for each disorder screening test; and~~

20 ~~(vi) authentication achieving potentiality of high~~
21 ~~throughput standards for statewide volume of each disorder~~
22 ~~screening test concomitant with each milestone described~~
23 ~~in items (i) through (iv) of this subsection (a 6).~~

24 ~~It is the goal of Public Act 97-532 that the expanded~~
25 ~~screening for the specified Lysosomal Storage Disorders begins~~
26 ~~within 2 years after August 23, 2011 (the effective date of~~

1 ~~Public Act 97-532). The Department is authorized to implement~~
2 ~~an additional fee for the screening prior to beginning the~~
3 ~~testing in order to accumulate the resources for start-up and~~
4 ~~other costs associated with implementation of the screening and~~
5 ~~thereafter to support the costs associated with screening and~~
6 ~~follow up programs for the specified Lysosomal Storage~~
7 ~~Disorders.~~

8 (a-7) (Blank). ~~In accordance with the timetable specified~~
9 ~~in this subsection (a-7), provide all newborns with expanded~~
10 ~~screening tests for the presence of Severe Combined~~
11 ~~Immunodeficiency Disease (SCID). The testing shall begin~~
12 ~~within 12 months following the occurrence of all of the~~
13 ~~following:~~

14 ~~(i) the establishment and verification of relevant and~~
15 ~~appropriate performance specifications as defined under~~
16 ~~the federal Clinical Laboratory Improvement Amendments and~~
17 ~~regulations thereunder for Federal Drug~~
18 ~~Administration cleared or in house developed methods,~~
19 ~~performed under an institutional review board approved~~
20 ~~protocol, if required;~~

21 ~~(ii) the availability of quality assurance testing and~~
22 ~~comparative threshold values for SCID;~~

23 ~~(iii) the acquisition and installment by the~~
24 ~~Department of the equipment necessary to implement the~~
25 ~~initial pilot and expanded statewide volume of screening~~
26 ~~tests for SCID;~~

1 ~~(iv) establishment of precise threshold values~~
2 ~~ensuring defined disorder identification for SCID;~~

3 ~~(v) authentication of pilot testing achieving each~~
4 ~~milestone described in items (i) through (iv) of this~~
5 ~~subsection (a 7) for SCID; and~~

6 ~~(vi) authentication achieving potentiality of high~~
7 ~~throughput standards for statewide volume of the SCID~~
8 ~~screening test concomitant with each milestone described~~
9 ~~in items (i) through (iv) of this subsection (a 7).~~

10 ~~It is the goal of Public Act 97-532 that the expanded~~
11 ~~screening for Severe Combined Immunodeficiency Disease begins~~
12 ~~within 2 years after August 23, 2011 (the effective date of~~
13 ~~Public Act 97-532). The Department is authorized to implement~~
14 ~~an additional fee for the screening prior to beginning the~~
15 ~~testing in order to accumulate the resources for start up and~~
16 ~~other costs associated with implementation of the screening and~~
17 ~~thereafter to support the costs associated with screening and~~
18 ~~follow up programs for Severe Combined Immunodeficiency~~
19 ~~Disease.~~

20 (a-8) (Blank). ~~In accordance with the timetable specified~~
21 ~~in this subsection (a 8), provide all newborns with expanded~~
22 ~~screening tests for the presence of certain Lysosomal Storage~~
23 ~~Disorders known as Mucopolysaccharidosis I (Hurlers) and~~
24 ~~Mucopolysaccharidosis II (Hunters). The testing shall begin~~
25 ~~within 12 months following the occurrence of all of the~~
26 ~~following:~~

1 ~~(i) the establishment and verification of relevant and~~
2 ~~appropriate performance specifications as defined under~~
3 ~~the federal Clinical Laboratory Improvement Amendments and~~
4 ~~regulations thereunder for Federal Drug~~
5 ~~Administration cleared or in house developed methods,~~
6 ~~performed under an institutional review board approved~~
7 ~~protocol, if required;~~

8 ~~(ii) the availability of quality assurance testing and~~
9 ~~comparative threshold values for each screening test and~~
10 ~~accompanying disorder;~~

11 ~~(iii) the acquisition and installment by the~~
12 ~~Department of the equipment necessary to implement the~~
13 ~~initial pilot and expanded statewide volume of screening~~
14 ~~tests for each disorder;~~

15 ~~(iv) establishment of precise threshold values~~
16 ~~ensuring defined disorder identification for each~~
17 ~~screening test;~~

18 ~~(v) authentication of pilot testing achieving each~~
19 ~~milestone described in items (i) through (iv) of this~~
20 ~~subsection (a-8) for each disorder screening test; and~~

21 ~~(vi) authentication achieving potentiality of high~~
22 ~~throughput standards for statewide volume of each disorder~~
23 ~~screening test concomitant with each milestone described~~
24 ~~in items (i) through (iv) of this subsection (a-8).~~

25 ~~It is the goal of Public Act 97-532 that the expanded~~
26 ~~screening for the specified Lysosomal Storage Disorders begins~~

1 ~~within 3 years after August 23, 2011 (the effective date of~~
2 ~~Public Act 97-532). The Department is authorized to implement~~
3 ~~an additional fee for the screening prior to beginning the~~
4 ~~testing in order to accumulate the resources for start-up and~~
5 ~~other costs associated with implementation of the screening and~~
6 ~~thereafter to support the costs associated with screening and~~
7 ~~follow up programs for the specified Lysosomal Storage~~
8 ~~Disorders.~~

9 (b) (Blank). ~~Maintain a registry of cases including~~
10 ~~information of importance for the purpose of follow up services~~
11 ~~to prevent intellectual disabilities.~~

12 (c) (Blank). ~~Supply the necessary metabolic treatment~~
13 ~~formulas where practicable for diagnosed cases of amino acid~~
14 ~~metabolism disorders, including phenylketonuria, organic acid~~
15 ~~disorders, and fatty acid oxidation disorders for as long as~~
16 ~~medically indicated, when the product is not available through~~
17 ~~other State agencies.~~

18 (d) (Blank). ~~Arrange for or provide public health nursing,~~
19 ~~nutrition and social services and clinical consultation as~~
20 ~~indicated.~~

21 (e) (Blank). ~~Require that all specimens collected pursuant~~
22 ~~to this Act or the rules and regulations promulgated hereunder~~
23 ~~be submitted for testing to the nearest Department of Public~~
24 ~~Health laboratory designated to perform such tests. The~~
25 ~~Department may develop a reasonable fee structure and may levy~~
26 ~~fees according to such structure to cover the cost of providing~~

~~this testing service. Fees collected from the provision of this testing service shall be placed in a special fund in the State Treasury, hereafter known as the Metabolic Screening and Treatment Fund. Other State and federal funds for expenses related to metabolic screening, follow up and treatment programs may also be placed in such Fund. Moneys shall be appropriated from such Fund to the Department of Public Health solely for the purposes of providing metabolic screening, follow up and treatment programs. Nothing in this Act shall be construed to prohibit any licensed medical facility from collecting additional specimens for testing for metabolic or neonatal diseases or any other diseases or conditions, as it deems fit. Any person violating the provisions of this subsection (c) is guilty of a petty offense.~~

(Source: P.A. 97-227, eff. 1-1-12; 97-532, eff. 8-23-11; 97-813, eff. 7-13-12.)

(410 ILCS 240/3.1 new)

Sec. 3.1. Lysosomal storage disorders. In accordance with the timetable specified in this Section, the Department shall provide all newborns with screening tests for the presence of certain lysosomal storage disorders known as Krabbe, Pompe, Gaucher, Fabry, and Niemann-Pick. The testing shall begin within 6 months following the occurrence of all of the following:

(1) the establishment and verification of relevant and

1 appropriate performance specifications as defined under
2 the federal Clinical Laboratory Improvement Amendments and
3 regulations thereunder for Federal Drug
4 Administration-cleared or in-house developed methods,
5 performed under an institutional review board approved
6 protocol, if required;

7 (2) the availability of quality assurance testing
8 methodology for these processes;

9 (3) the acquisition and installment by the Department
10 of the equipment necessary to implement the screening
11 tests;

12 (4) the establishment of precise threshold values
13 ensuring defined disorder identification for each
14 screening test;

15 (5) the authentication of pilot testing achieving each
16 milestone described in items (1) through (4) of this
17 Section for each disorder screening test; and

18 (6) the authentication of achieving the potential of
19 high throughput standards for statewide volume of each
20 disorder screening test concomitant with each milestone
21 described in items (1) through (4) of this Section.

22 It was the goal of Public Act 97-532 that the screening for
23 the specified lysosomal storage disorders begins within 2 years
24 after August 23, 2011 (the effective date of Public Act
25 97-532). The Department is authorized to implement an
26 additional fee for the screening prior to beginning the testing

1 in order to accumulate the resources for start-up and other
2 costs associated with implementation of the screening and
3 thereafter to support the costs associated with screening and
4 follow-up programs for the specified lysosomal storage
5 disorders.

6 (410 ILCS 240/3.2 new)

7 Sec. 3.2. Severe combined immunodeficiency disease. In
8 accordance with the timetable specified in this Section, the
9 Department shall provide all newborns with screening tests for
10 the presence of severe combined immunodeficiency disease
11 (SCID). The testing shall begin within 12 months following the
12 occurrence of all of the following:

13 (1) the establishment and verification of relevant and
14 appropriate performance specifications as defined under
15 the federal Clinical Laboratory Improvement Amendments and
16 regulations thereunder for Federal Drug
17 Administration-cleared or in-house developed methods,
18 performed under an institutional review board approved
19 protocol, if required;

20 (2) the availability of quality assurance testing and
21 comparative threshold values for SCID;

22 (3) the acquisition and installment by the Department
23 of the equipment necessary to implement the initial pilot
24 and statewide volume of screening tests for SCID;

25 (4) the establishment of precise threshold values

1 ensuring defined disorder identification for SCID;

2 (5) the authentication of pilot testing achieving each
3 milestone described in items (1) through (4) of this
4 Section for SCID; and

5 (6) the authentication of achieving the potential of
6 high throughput standards for statewide volume of the SCID
7 screening test concomitant with each milestone described
8 in items (1) through (4) of this Section.

9 It was the goal of Public Act 97-532 that the screening for
10 severe combined immunodeficiency disease begins within 2 years
11 after August 23, 2011 (the effective date of Public Act
12 97-532). The Department is authorized to implement an
13 additional fee for the screening prior to beginning the testing
14 in order to accumulate the resources for start-up and other
15 costs associated with implementation of the screening and
16 thereafter to support the costs associated with screening and
17 follow-up programs for severe combined immunodeficiency
18 disease.

19 (410 ILCS 240/3.3 new)

20 Sec. 3.3. Mucopolysaccharidosis disorders. In accordance
21 with the timetable specified in this Section, the Department
22 shall provide all newborns with screening tests for the
23 presence of certain lysosomal storage disorders known as
24 mucopolysaccharidosis I (Hurlers) and mucopolysaccharidosis II
25 (Hunters). The testing shall begin within 12 months following

1 the occurrence of all of the following:

2 (1) the establishment and verification of relevant and
3 appropriate performance specifications as defined under
4 the federal Clinical Laboratory Improvement Amendments and
5 regulations thereunder for Federal Drug
6 Administration-cleared or in-house developed methods,
7 performed under an institutional review board approved
8 protocol, if required;

9 (2) the availability of quality assurance testing and
10 comparative threshold values for each screening test and
11 accompanying disorder;

12 (3) the acquisition and installment by the Department
13 of the equipment necessary to implement the initial pilot
14 and statewide volume of screening tests for each disorder;

15 (4) the establishment of precise threshold values
16 ensuring defined disorder identification for each
17 screening test;

18 (5) the authentication of pilot testing achieving each
19 milestone described in items (1) through (4) of this
20 Section for each disorder screening test; and

21 (6) the authentication of achieving the potential of
22 high throughput standards for statewide volume of each
23 disorder screening test concomitant with each milestone
24 described in items (1) through (4) of this Section.

25 It was the goal of Public Act 97-532 that the screening for
26 the specified lysosomal storage disorders begins within 3 years

1 after August 23, 2011 (the effective date of Public Act
2 97-532). The Department is authorized to implement an
3 additional fee for the screening prior to beginning the testing
4 in order to accumulate the resources for start-up and other
5 costs associated with implementation of the screening and
6 thereafter to support the costs associated with screening and
7 follow-up programs for the specified lysosomal storage
8 disorders.

9 Section 10. The Genetic and Metabolic Diseases Advisory
10 Committee Act is amended by changing Section 5 as follows:

11 (410 ILCS 265/5)

12 Sec. 5. Genetic and Metabolic Diseases Advisory Committee.

13 (a) The Director of Public Health shall create the Genetic
14 and Metabolic Diseases Advisory Committee to advise the
15 Department of Public Health regarding issues relevant to
16 newborn screenings of metabolic diseases.

17 (b) The purposes of Metabolic Diseases Advisory Committee
18 are all of the following:

19 (1) Advise the Department regarding issues relevant to
20 its Genetics Program.

21 (2) Advise the Department regarding optimal laboratory
22 methodologies for screening of the targeted conditions.

23 (3) Recommend to the Department consultants who are
24 qualified to diagnose a condition detected by screening,

1 provide management of care, and genetic counseling for the
2 family.

3 (4) Monitor the incidence of each condition for which
4 newborn screening is done, evaluate the effects of
5 treatment and genetic counseling, and provide advice on
6 disorders to be included in newborn screening panel.

7 (5) Advise the Department on educational programs for
8 professionals and the general public.

9 (6) Advise the Department on new developments and areas
10 of interest in relation to the Genetics Program.

11 (7) Any other matter deemed appropriate by the
12 Committee and the Director.

13 (c) The Committee shall consist of 20 members appointed by
14 the Director of Public Health. Membership shall include
15 physicians, geneticists, nurses, nutritionists, and other
16 allied health professionals, as well as patients and parents.
17 Ex-officio members may be appointed, but shall not have voting
18 privileges.

19 (d) Members of the Committee may receive compensation for
20 necessary expenses incurred in the performance of their duties.

21 (Source: P.A. 95-695, eff. 11-5-07.)

22 Section 99. Effective date. This Act takes effect upon
23 becoming law.