

NEWBORN CHILD TESTING ACT - NEWBORN CHILD SCREENING AND
FOLLOW-UP PROGRAM AND MANDATED SCREENING AND FOLLOW-UP

Act of Nov. 25, 2020, P.L. , No. 133

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No. 2020-133

SB 983

AN ACT

Amending the act of September 9, 1965 (P.L.497, No.251), entitled "An act requiring physicians, hospitals and other institutions to administer or cause to be administered tests for genetic diseases upon infants in certain cases," further providing for definitions and for Newborn Child Screening and Follow-up Program; and providing for mandated screening and follow-up.

The General Assembly of the Commonwealth of Pennsylvania hereby enacts as follows:

Section 1. The definitions of "board" and "disease" in section 2 of the act of September 9, 1965 (P.L.497, No.251), known as the Newborn Child Testing Act, are amended and the section is amended by adding definitions to read:

Section 2. Definitions.--The following words and phrases when used in this act shall have the meanings given to them in this section unless the context clearly indicates otherwise:

"Birthing facilities." An inpatient or ambulatory health care facility licensed by the department that provides birthing and newborn care services.

"Board." The [State Advisory Health] Newborn Screening and Follow-up Technical Advisory Board in the Department of Health.

"Certified nurse-midwife." An individual licensed by the State Board of Medicine to practice midwifery under section 35 of the act of December 20, 1985 (P.L.457, No.112), known as the Medical Practice Act of 1985.

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"Disease." Diseases listed by the Department of Health by regulation which lead to [mental retardation or physical defects] intellectual disability, physical disability or death, including, without limitation, Phenylketonuria (PKU), maple syrup urine disease (MSUD) and sickle-cell disease (hemoglobinopathies).

"Health care practitioner." As the term is defined in section 103 of the act of July 19, 1979 (P.L.130, No.48), known as the Health Care Facilities Act.

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"Secretary." The Secretary of Health of the Commonwealth.

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"Unlicensed midwife." The term includes nationally certified midwives, traditional midwives, Amish, Mennonite or Plain midwives and other specific cultural or spiritual community-based midwives not licensed to practice midwifery in this Commonwealth.

Section 2. Section 3(a), (b.1), (d) and (e) of the act are amended and the section is amended by adding a subsection to read:

Section 3. Newborn Child Screening and Follow-up Program.--(a) In order to assist health care providers to determine whether treatment or other services are necessary to

avert [mental retardation, permanent disabilities] **intellectual disability, physical disability** or death, the department, with the approval of the Newborn Screening and Follow-up Technical Advisory [Committee] **Board**, shall establish a program providing for:

(1) The screening tests of newborn children **and follow-up services** for the following diseases:

- (i) Phenylketonuria (PKU).
- (ii) Maple syrup urine disease (MSUD).
- (iii) Sickle-cell disease (hemoglobinopathies).
- (iv) Galactosemia.
- (v) Congenital adrenal hyperplasia (CAH).
- (vi) Primary congenital hypothyroidism.
- [(vii) Certain Lysosomal storage disorders (LSDs),

including:

- (A) Globoid Cell Leukodystrophy (Krabbe).
- (B) Fabry.
- (C) Pompe.
- (D) Niemann-Pick.
- (E) Gaucher.
- (F) Hurler Syndrome (MPS I).

(2) Follow-up services relating to case management, referrals, confirmatory testing, assessment and diagnosis of newborn children with abnormal, inconclusive or unacceptable screening test results for the following diseases:

- (i) Phenylketonuria (PKU).
- (ii) Maple syrup urine disease (MSUD).
- (iii) Sickle-cell disease (hemoglobinopathies).
- (iv) Isovaleric acidemia/Isovalery-CoA dehydrogenase deficiency (IVA).
- (v) Glutaric acidemia Type I/Glutaryl-CoA dehydrogenase deficiency Type I (GA I).
- (vi) 3-Hydroxy 3-methylglutaryl-CoA lyase deficiency (HMG).
- (vii) Multiple carboxylase deficiency (MCD).
- (viii) Methylmalonic acidemia (mutase deficiency) (MUT).
- (ix) Methylmalonic acidemia (Cbl A,B).
- (x) 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC).
- (xi) Propionic acidemia/Propionyl-CoA carboxylase deficiency (PROP).
- (xii) Beta-ketothiolase deficiency (BKT).
- (xiii) Medium chain acyl-CoA dehydrogenase deficiency (MCAD).
- (xiv) Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD).
- (xv) Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD).
- (xvi) Trifunctional protein deficiency (TFP).
- (xvii) Carnitine uptake defect (CUD).
- (xviii) Homocystinuria (HCY).
- (xix) Tyrosinemia type I (TYR I).
- (xx) Argininosuccinic acidemia (ASA).
- (xxi) Citrullinemia (CIT).
- (xxii) Hb S/Beta-thalassemia (Hb S/Th).
- (xxiii) Hb S/C disease (Hb S/C).
- (xxiv) Congenital hypothyroidism (HYPOTH).
- (xxv) Biotinidase deficiency (BIOT).
- (xxvi) Congenital adrenal hyperplasia (CAH).
- (xxvii) Galactosemia (GALT).
- (xxviii) Cystic fibrosis (CF).]
- (vii) Pompe.**
- (viii) Hurler Syndrome (MPS I).**
- (ix) Adrenoleukodystrophy (ALD).**

(x) Spinal Muscular Atrophy (SMA).

(2) A birthing facility, certified nurse-midwife, unlicensed midwife or health care practitioner shall order the screening tests of newborn children for the following diseases:

(i) Isovaleric acidemia/Isovalery-CoA dehydrogenase deficiency (IVA).

(ii) Glutaric acidemia Type I/Glutaryl-CoA dehydrogenase deficiency Type I (GA I).

(iii) 3-Hydroxy 3-methylglutaryl-CoA lyase deficiency (HMG).

(iv) Multiple carboxylase deficiency (MCD).

(v) Methylmalonic acidemia (mutase deficiency) (MUT).

(vi) Methylmalonic acidemia (Cbl A, B).

(vii) 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC).

(viii) Propionic acidemia/Propionyl-CoA carboxylase deficiency (PROP).

(ix) Beta-ketothiolase deficiency (BKT).

(x) Medium chain acyl-CoA dehydrogenase deficiency (MCAD).

(xi) Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD).

(xii) Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD).

(xiii) Trifunctional protein deficiency (TFP).

(xiv) Carnitine uptake defect (CUD).

(xv) Homocystinuria (HCY).

(xvi) Tyrosinemia type I (TYR I).

(xvii) Argininosuccinic acidemia (ASA).

(xviii) Citrullinemia (CIT).

(xix) Hb S/Beta-thalassemia (Hb S/Th).

(xx) Hb S/C disease (Hb S/C).

(xxi) Biotinidase deficiency (BIOT).

(xxii) Cystic fibrosis (CF).

(xxiii) Severe combined immunodeficiency disease (SCID).

(xxiv) Globoid Cell Leukodystrophy (Krabbe).

(b.1) All laboratories performing the screening tests for newborn children shall report the results to the department for follow-up activities. The department shall provide follow-up services through the program relating to case management, referrals, confirmatory testing, assessment and diagnosis of newborn children with abnormal, inconclusive or unacceptable screening test results up to a newborn child's first year of life.

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(d) The department, with the approval of the Newborn Screening and Follow-up Technical Advisory [Committee] Board, shall establish, by transmitting notice to the Legislative Reference Bureau for periodic publication in the Pennsylvania Bulletin, changes to the lists under subsection (a)(1) and (2) of those diseases for which newborn children shall be screened and laboratory screening results reported. Prior to making any change, the department and board shall jointly transmit a notice to the Legislative Reference Bureau for publication in the Pennsylvania Bulletin that establishes a public comment period of at least 30 days.

(d.1) The board shall consist of multidisciplinary members appointed by the secretary. The board shall include at least the following:

(1) An ethicist.

(2) Three pediatric physicians.

(3) A neonatologist.

(4) A genetic counselor.

(5) A hematologist.

(6) Two clinical geneticists.

- (7) A nurse-midwife.
- (8) A parent advocate.
- (9) A representative recommended by a hospital association.

(e) Notwithstanding any provisions of this act or the act of April 23, 1956 (1955 P.L.1510, No.500), known as the "Disease Prevention and Control Law of 1955," to the contrary, test results and diagnoses based upon screening tests for the diseases listed in this section for newborn children shall be reported to the department. The department shall establish, by **transmitting notice to the Legislative Reference Bureau for** periodic publication in the Pennsylvania Bulletin, the method for reporting test results to the department.

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Section 3. The act is amended by adding a section to read:

Section 3.1. Mandated Screening and Follow-up.--Diseases and conditions mandated for screenings and follow-up services shall, at a minimum, include:

- (1) diseases listed under section 3(a);
- (2) diseases added by the board under section 3(d) to the list of diseases under section 3(a); and
- (3) conditions listed in the Recommended Uniform Screening Panel by the United States Department of Health and Human Services.

Section 4. This act shall take effect in 180 days.

APPROVED--The 25th day of November, A.D. 2020.

TOM WOLF