35 P.S. § 621, et. seq.

§ 621. Short title

This act shall be known and may be cited as the "Newborn Child Testing Act."

§ 622. 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:

"BOARD." The State Advisory Health Board in the Department of Health.

"DEPARTMENT." The Department of Health of the Commonwealth.

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

"HEALTH CARE PROVIDER." A health care facility or health care practitioner as defined by regulations of the Department of Health.

"NEWBORN CHILD." A child less than 28 days of age.

"PROGRAM." The Newborn Child Screening and Follow-up Program administered by the Department of Health.

"REPEAT SPECIMEN." A second or subsequent blood specimen collected from a newborn child for the same purpose.

"UNACCEPTABLE SPECIMEN." A newborn child's blood specimen which is not suitable in quality or quantity to perform newborn screening or confirmatory testing for one or more of the diseases covered by this act or the regulations promulgated thereunder.

Section 3, prior to July 1, 2009:

§ 623. 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 or death, the department, with the approval of the board, shall establish a program providing for:
 - (1) The screening tests of newborn children for diseases.
- (2) Follow-up services relating to confirmatory testing, assessment and diagnosis of newborn children with abnormal or inconclusive screening test results.
- (b) The department, with the approval of the board, shall establish by regulation those diseases, in addition to phenylketonuria (PKU), maple syrup urine disease (MSUD) and sickle-cell disease

(hemoglobinopathies), for which newborn children shall be tested and the methods for testing and disseminating test results.

(c) No screening test shall be performed if a parent or guardian dissents on the ground that the test conflicts with a religious belief or practice.

Section 3 effective July 1, 2009; amended by Act 36 of 2008:

§ 623. 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 or death, the department, with the approval of the Newborn Screening and Follow-up Technical Advisory Board, shall establish a program providing for:
 - (1) The screening tests of newborn children 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.
- (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-Methylcrontonyl-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).
- (b) Deleted effective July 1, 2009.
- (b.1) All laboratories performing the screening tests for newborn children shall report the results to the department for follow-up activities.
- (c) No screening test shall be performed if a parent or guardian dissents on the ground that the test conflicts with a religious belief or practice.
- (d) The department, with the approval of the Newborn Screening and Follow-up Technical Advisory Board, shall establish, by 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.
- (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 periodic publication in the Pennsylvania Bulletin, the method for reporting test results to the department.
 - (f) Test results for genetic diseases listed in this section and any diseases subsequently added

by the department under subsection (d) shall be subject to the confidentiality provisions of the "Disease Prevention and Control Law of 1955."

§ 624. Procurement of specimens by health care providers

- (a) Health care providers shall cause to be procured blood specimens of newborn children for required screening and confirmatory tests and send such specimens to a testing laboratory designated by the department.
- (b) If the initial specimen is an unacceptable specimen or as otherwise required by the department by regulation, the health care provider shall collect a repeat specimen for screening and confirmatory tests.

§ 625. Regulations

The department, with the approval of the board, shall have the authority to promulgate regulations for the implementation and administration of this act.