PRIOR PRINTER'S NO. 1045 PRINTER'S NO. 1420

THE GENERAL ASSEMBLY OF PENNSYLVANIA

SENATE BILL

No. 819 Session of 2005

INTRODUCED BY ORIE, KITCHEN, ERICKSON, BROWNE, PILEGGI, TARTAGLIONE, COSTA, PIPPY, WONDERLING, C. WILLIAMS, RHOADES, O'PAKE AND RAFFERTY, JUNE 30, 2005

SENATOR CORMAN, PUBLIC HEALTH AND WELFARE, AS AMENDED, DECEMBER 13, 2005

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 phenylketonuria and other metabolic diseases upon infants in certain cases," further providing for newborn child screening and testing; and making editorial changes.

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

Section 1. The title of the act of September 9, 1965 (P.L.497, No.251), known as the Newborn Child Testing Act, is amended to read:

Requiring physicians, hospitals and other institutions to administer or cause to be administered SCREENING AND tests for [phenylketonuria and other metabolic] genetic, METABOLIC, HORMONAL AND FUNCTIONAL CONDITIONS OR diseases upon infants in certain cases.

Section 2. Section 3 of the act, added July 9, 1992 (P.L.398, No.86), is amended 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 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**]** CONDITIONS.

(2) Follow-up services relating to confirmatory SCREENING AND 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 SCREENED AND tested and the methods for ~~testing [and disseminating~~ SCREENING AND TESTING AND DISSEMINATING SCREENING AND test results.~~] shall at a minimum include the following:~~

~~(1) Argininosuccinic acidemia (ASA).~~

~~(2) Beta-ketothiolase deficiency (BKT).~~

~~(3) Biotinidase deficiency (BIOT).~~

~~(4) Carnitine uptake defect (CUD).~~

~~(5) Citrullinemia (CIT).~~

~~(6) Congenital adrenal hyperplasia (CAH).~~

~~(7) Congenital hypothyroidism (HYPOTH).~~

~~(8) Cystic fibrosis (CF).~~

~~(9) Galactosemia (GALT).~~

~~(10) Glutaric acidemia type I (GAI)~~

~~(11) Hb S/Beta-thalassemia (Hb S/Th).~~

~~(12) Hb S/C disease (Hb S/C).~~

~~(13) Hearing deficiency (HEAR).~~

~~(14) Homocystinuria (HCY).~~

~~(15) Isovaleric acidemia (IVA).~~

~~(16) Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD).~~

~~(17) Maple syrup urine disease (MSUD).~~

~~(18) Medium-chain acyl-CoA dehydrogenase deficiency (MCAD).~~

~~(19) Methylmalonic acidemia (Cbl A,B).~~

~~(20) Methylmalonic acidemia (mutase deficiency) (MUT).~~

~~(21) 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC).~~

~~(22) Multiple carboxylase deficiency (MCD).~~

~~(23) 3-OH 3-CH3 glutaric aciduria (HMG).~~

~~(24) Phenylketonuria (PKU).~~

~~(25) Propionic acidemia (PROP).~~

~~(26) Sickle cell anemia (SCA).~~

~~(27) Trifunctional protein deficiency (TFP).~~

~~(28) Tyrosinemia type I (TYRI).~~

~~(29) Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD).~~

~~The following conditions should be included as "report only":~~

~~(1) Argininemia (ARG).~~

~~(2) Carnitine acylcarnitine translocase deficiency (CACT).~~

~~(3) Carnitine palmitoyltransferase I deficiency (liver) (CPT-IA).~~

~~(4) Carnitine palmitoyltransferase II deficiency (CPT-II).~~

~~(5) Citrullinemia type 2 (CIT-II).~~

~~(6) Defects of biopterin cofactor biosynthesis (BIOPT (BS)).~~

~~(7) Defects of biopterin cofactor regeneration (BIOPT (REG)).~~

~~(8) Dienoyl-CoA reductase deficiency (DE REDUCT).~~

~~(9) Galactokinase deficiency (GALK).~~

~~(10) Galactose epimerase deficiency (GALE).~~

~~(11) Glutaric acidemia type 2 (GA2).~~

~~(12) Hemoglobin variants (including Hb E=Hemoglobin E) (Variant Hb).~~

~~(13) Hypermethioninemia (MET).~~

~~(14) Hyperphenylalaninemia, benign (not PKU) (Hyper-PHE).~~

~~(15) Isobutyryl-CoA dehydrogenase deficiency (IBG).~~

~~(16) Malonic acidemia (MAL).~~

~~(17) Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT).~~

~~(18) Medium/short chain L-3-OH acyl-CoA dehydrogenase deficiency (M/SCHAD).~~

~~(19) 2-Methyl 3-hydroxy butyric aciduria (2M3HBA).~~

~~(20) 2-Methybutyryl-CoA dehydrogenase deficiency (2MBG).~~

~~(21) 3-Methylglutaconic aciduria (3MGA).~~

~~(22) Methylmalonic acidemia, Cbl C and Cbl D forms (Cbl C,D).~~

~~(23) Short chain acyl-CoA dehydrogenase deficiency (SCAD).~~

~~(24) Tyrosinemia type 2 (TYR-II).~~

~~(25) Tyrosinemia3 type 3 (TYR-III).~~

~~(b.1) The listing of tests for heritable disorders may be revised to include conditions as deemed appropriate by the department based on the recommendations of the American College of Medical Genetics.~~

(B.1) THE DEPARTMENT SHALL PROVIDE FOLLOW-UP SERVICES <——
RELATING TO CONFIRMATORY SCREENING AND TESTING, ASSESSMENT AND DIAGNOSIS OF NEWBORN CHILDREN WITH ABNORMAL OR INCONCLUSIVE SCREENING TEST RESULTS FOR THE FOLLOWING DISEASES:

(1) PHENYLKETONURIA (PKU).

(2) MAPLE SYRUP URINE DISEASE (MSUD).

(3) SICKLE-CELL DISEASE (HEMOGLOBINOPATHIES).

(4) ISOVALERIC ACIDEMIA/ISOVALERY-COA DEHYDROGENASE DEFICIENCY (IVA).

(5) GLUTARIC ACIDEMIA TYPE I/GLUTARYL-COA DEHYDROGENASE DEFICIENCY TYPE I (GA I).

(6) 3-RYDROXY 3-METHYLGLUTARYL-COA LYASE DEFICIENCY (HMG).

(7) MULTIPLE CARBOXYLASE DEFICIENCY (MCD).

(8) METHYLMALONIC ACIDEMIA (MUTASE DEFICIENCY) (MUI).

(9) METHYLMALONIC ACIDEMIA (CBL A,B).

(10) 3-METHYLCRONTONYL-COA CARBOXYLASE DEFICIENCY (3MCC).

(11) PROPIONIC ACIDEMIA/PROPIONYL-COA CARBOXYLASE DEFICIENCY (PROP).

(12) BETA-KETOTHIOLASE DEFICIENCY (BKT).

(13) MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (MCAD).

(14) VERY LONG-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (VLCAD).

(15) LONG-CHAIN L-3-OH ACYL-COA DEHYDROGENASE DEFICIENCY (LCHAD).

(16) TRIFUNCTIONAL PROTEIN DEFICIENCY (TFP).

(17) CARNITINE UPTAKE DEFECT (CUD).

(18) HOMOCYSTINURIA (HCY).

(19) TYROSINEMIA TYPE I (TYR I).

(20) ARGININOSUCCINIC ACIDEMIA (ASA).

(21) CITRULLINEMIA (CIT).

(22) HB S/BETA-THALASSEMIA (HB S/TH).

(23) HB S/C DISEASE (HB S/C).

(24) GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY (G6PD).

(25) CONGENITAL HYPOTHYROIDISM (HYPDTR).

(26) BIOTINIDASE DEFICIENCY (BIOT).

(27) CONGENITAL ADRENAL HYPERPLASIA (CAH).

(28) GALACTOSEMIA (GALT).

(29) CYSTIC FIBROSIS (CF).

(B.2) ALL LABORATORIES PERFORMING THE SCREENING TESTS FOR NEWBORN CHILDREN SHALL REPORT ANY ABNORMAL OR INCONCLUSIVE 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 sum of $2,000,000, or as much thereof as may be necessary, is hereby appropriated to the Department of Health to carry out the purpose of this act.~~ <—

Section 3. This act shall take effect in 60 days.