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THE GENERAL ASSEMBLY OF PENNSYLVANIA

HOUSE BILL

No. 2039 Session of 2018

INTRODUCED BY CHARLTON, SCHLOSSBERG, CALTAGIRONE, DRISCOLL, O'NEILL, SOLOMON, BARRAR, MILLARD, ROEBUCK, DiGIROLAMO, GILLEN, WARREN AND WATSON, JANUARY 25, 2018

REFERRED TO COMMITTEE ON HEALTH, JANUARY 25, 2018

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 newborn child screening and follow-up programs.

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

Section 1. Section 3(a) of the act of September 9, 1965 (P.L.497, No.251), known as the Newborn Child Testing Act, 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 Newborn Screening and Follow-up Technical Advisory Committee, 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.

(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).

(G) Spinal muscular atrophy (SMA).

(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).

(xxix) Spinal muscular atrophy (SMA).

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Section 2. This act shall take effect in 60 days.