
THE GENERAL ASSEMBLY OF PENNSYLVANIA

HOUSE BILL

No. 2484 Session of
2018

INTRODUCED BY M. QUINN, KINSEY, MILLARD, READSHAW, HELM, THOMAS,
LONGIETTI, BERNSTINE, J. McNEILL, RYAN, DeLUCA, WATSON AND
GILLEN, JUNE 12, 2018

REFERRED TO COMMITTEE ON HEALTH, JUNE 12, 2018

AN ACT

1 Amending the act of September 9, 1965 (P.L.497, No.251),
2 entitled, "An act requiring physicians, hospitals and other
3 institutions to administer or cause to be administered tests
4 for genetic diseases upon infants in certain cases," further
5 providing for newborn child screening and follow-up program.

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

8 Section 1. Section 3(a) of the act of September 9, 1965
9 (P.L.497, No.251), known as the Newborn Child Testing Act, is
10 amended to read:

11 Section 3. Newborn Child Screening and Follow-up Program.--

12 (a) In order to assist health care providers to determine
13 whether treatment or other services are necessary to avert
14 mental retardation, permanent disabilities or death, the
15 department, with the approval of the Newborn Screening and
16 Follow-up Technical Advisory Committee, shall establish a
17 program providing for:

18 (1) The screening tests of newborn children for the
19 following diseases:

- 1 (i) Phenylketonuria (PKU).
- 2 (ii) Maple syrup urine disease (MSUD).
- 3 (iii) Sickle-cell disease (hemoglobinopathies).
- 4 (iv) Galactosemia.
- 5 (v) Congenital adrenal hyperplasia (CAH).
- 6 (vi) Primary congenital hypothyroidism.
- 7 (vii) Certain Lysosomal storage disorders (LSDs), including:
 - 8 (A) Globoid Cell Leukodystrophy (Krabbe).
 - 9 (B) Fabry.
 - 10 (C) Pompe.
 - 11 (D) Niemann-Pick.
 - 12 (E) Gaucher.
 - 13 (F) Hurler Syndrome (MPS I).
- 14 (viii) Spinal muscular atrophy (SMA).

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

- 19 (i) Phenylketonuria (PKU).
- 20 (ii) Maple syrup urine disease (MSUD).
- 21 (iii) Sickle-cell disease (hemoglobinopathies).
- 22 (iv) Isovaleric acidemia/Isovalery-CoA dehydrogenase
23 deficiency (IVA).
- 24 (v) Glutaric acidemia Type I/Glutaryl-CoA dehydrogenase
25 deficiency Type I (GA I).
- 26 (vi) 3-Hydroxy 3-methylglutaryl-CoA lyase deficiency (HMG).
- 27 (vii) Multiple carboxylase deficiency (MCD).
- 28 (viii) Methylmalonic acidemia (mutase deficiency) (MUT).
- 29 (ix) Methylmalonic acidemia (Cbl A,B).
- 30 (x) 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC).

- 1 (xi) Propionic acidemia/Propionyl-CoA carboxylase deficiency
2 (PROP).
- 3 (xii) Beta-ketothiolase deficiency (BKT).
- 4 (xiii) Medium chain acyl-CoA dehydrogenase deficiency
5 (MCAD).
- 6 (xiv) Very long-chain acyl-CoA dehydrogenase deficiency
7 (VLCAD).
- 8 (xv) Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
9 (LCHAD).
- 10 (xvi) Trifunctional protein deficiency (TFP).
- 11 (xvii) Carnitine uptake defect (CUD).
- 12 (xviii) Homocystinuria (HCY).
- 13 (xix) Tyrosinemia type I (TYR I).
- 14 (xx) Argininosuccinic acidemia (ASA).
- 15 (xxi) Citrullinemia (CIT).
- 16 (xxii) Hb S/Beta-thalassemia (Hb S/Th).
- 17 (xxiii) Hb S/C disease (Hb S/C).
- 18 (xxiv) Congenital hypothyroidism (HYPOTH).
- 19 (xxv) Biotinidase deficiency (BIOT).
- 20 (xxvi) Congenital adrenal hyperplasia (CAH).
- 21 (xxvii) Galactosemia (GALT).
- 22 (xxviii) Cystic fibrosis (CF).
- 23 (xxix) Spinal muscular atrophy (SMA).

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