
THE GENERAL ASSEMBLY OF PENNSYLVANIA

HOUSE BILL

No. 2479 Session of
2014

INTRODUCED BY STERN, B. BOYLE, V. BROWN, BOBACK, MUNDY,
MIRABITO, KIM, BROWNLIEE, WATSON, MILLARD, DAY, BAKER, THOMAS,
O'BRIEN, GODSHALL, COHEN, MURT, FARINA, JAMES, KAUFFMAN,
SABATINA, READSHAW, CLYMER, HEFFLEY, HARHAI, PETRI AND
GINGRICH, SEPTEMBER 17, 2014

REFERRED TO COMMITTEE ON HUMAN SERVICES, SEPTEMBER 17, 2014

AN ACT

1 Amending the act of September 9, 1965 (P.L.497, No.251),
2 entitled, as amended, "An act requiring physicians, hospitals
3 and other institutions to administer or cause to be
4 administered tests for genetic diseases upon infants in
5 certain cases," further providing for the Newborn Child
6 Screening and Follow-up Program.

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

9 Section 1. Section 3(a)(2) of the act of September 9, 1965
10 (P.L.497, No.251), known as the Newborn Child Testing Act,
11 amended July 4, 2008 (P.L.288, No.36), is amended to read:

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

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

1 * * *

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

6 (i) Phenylketonuria (PKU).

7 (ii) Maple syrup urine disease (MSUD).

8 (iii) Sickle-cell disease (hemoglobinopathies).

9 (iv) Isovaleric acidemia/Isovalery-CoA dehydrogenase
10 deficiency (IVA).

11 (v) Glutaric acidemia Type I/Glutaryl-CoA dehydrogenase
12 deficiency Type I (GA I).

13 (vi) 3-Hydroxy 3-methylglutaryl-CoA lyase deficiency (HMG).

14 (vii) Multiple carboxylase deficiency (MCD).

15 (viii) Methylmalonic acidemia (mutase deficiency) (MUT).

16 (ix) Methylmalonic acidemia (Cbl A,B).

17 (x) 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC).

18 (xi) Propionic acidemia/Propionyl-CoA carboxylase deficiency
19 (PROP).

20 (xii) Beta-ketothiolase deficiency (BKT).

21 (xiii) Medium chain acyl-CoA dehydrogenase deficiency
22 (MCAD).

23 (xiv) Very long-chain acyl-CoA dehydrogenase deficiency
24 (VLCAD).

25 (xv) Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
26 (LCHAD).

27 (xvi) Trifunctional protein deficiency (TFP).

28 (xvii) Carnitine uptake defect (CUD).

29 (xviii) Homocystinuria (HCY).

30 (xix) Tyrosinemia type I (TYR I).

- 1 (xx) Argininosuccinic acidemia (ASA).
2 (xxi) Citrullinemia (CIT).
3 (xxii) Hb S/Beta-thalassemia (Hb S/Th).
4 (xxiii) Hb S/C disease (Hb S/C).
5 (xxiv) Congenital hypothyroidism (HYPOTH).
6 (xxv) Biotinidase deficiency (BIOT).
7 (xxvi) Congenital adrenal hyperplasia (CAH).
8 (xxvii) Galactosemia (GALT).
9 (xxviii) Cystic fibrosis (CF).
10 (xxix) Nonketotic hyperglycinemia (NKH).
11 * * *
12 Section 2. This act shall take effect in 60 days.