# Expanded Newborn Screening Comparative Table

Numbers = disorders are screened for and counted by testing lab.

Bullet (•) = disorders are screened for but not counted as a separate disorder by testing lab.

** Newborn hearing screening is also recommended, but is not included in our metabolic table.*

| Hawaii | California[1] | ACMG* | Baylor | Pediatrix[2] |
|---|---|---|---|---|
| **AAD:** | **AAD:** | **AAD:** | **AAD:** | **AAD:** |
| 1) ARG | 1) 5-oxoprolinuria | | 1) 5-oxoprolinuria | 1) 5-oxoprolinuria |
| | 2) ARG | | 2) ARG | 2) ARG |
| 2) ASAL | 3) ASAL | 1) ASAL | 3) ASAL | ASAL |
| | • Acute onset | | • Acute onset | 3) Acute onset |
| | • Chronic onset | | • Chronic onset | 4) Chronic onset |
| 3) ASAS | 4) ASAS | 2) ASAS | 4) ASAS | ASAS |
| • Acute onset | • Acute onset | | • Acute onset | 5) Acute onset |
| • Chronic onset | • Chronic onset | | • Chronic onset | 6) Chronic onset |
| • CIT-type 2 | 5) CIT-type 2 | | | |
| | | | | 7) CPS[3] |
| 4) HCY | 6) HCY | 3) HCY | 5) HCY | 8) HCY |
| • MAT | 7) MAT | 4) MAT | 6) MAT | 9) MAT |
| | 8) HHH | 5) HHH | 7) HHH | 10) HHH[3] |
| | | | | 11) HOGA[3] |
| 5) MSUD | 9) MSUD | 6) MSUD | 8) MSUD | MSUD |
| • Classical | • Classical | | • Classical | 12) Classical |
| • Intermediate | • Intermediate | | • Intermediate | 13) Intermediate |
| | 10) NKH | | 9) NKH | |
| 6) PKU | PKU | 7) PKU | 10) PKU | PKU |
| • Classical | 11) Classical | | • Classical | 14) Classical |
| • HPHE | 12) HPHE | | • HPHE | 15) HPHE |
| • Benign | • Benign | | • Benign | • Benign |
| • BPT (4 types)[4] | 13) BPT (4 types)[4] | | • BPT (4 types)[4] | 16) BPT (4 types)[4] |
| 7) TYR | 14) TYR | | TYR | TYR |
| • TYR-type 1 | • TYR-type 1 | 8) TYR-type 1 | 11) TYR-type 1 | 17) TYR-type 1 |
| • TYR-type 2 | • TYR-type 2 | | 12) TYR-type 2 | 18) TYR-type 2 |
| • TYR-type 3 | • TYR-type 3 | | | 19) TYR-type 3 |
| • TNTYR | • TNTYR | | | 20) TNTYR |
| **OAD:** | **OAD:** | **OAD:** | **OAD:** | **OAD:** |
| 8) 2MBC | 15) 2MBC | | 13) 2MBC | 21) 2MBC |
| 9) 3MCC | 16) 3MCC | 9) 3MCC | 14) 3MCC | 22) 3MCC |
| 10) 3MGH | 3MGH | | | 23) 3MGH |
| • 3MGH-type 1 | 17) 3MGH-type 1 | | | |
| 11) BKD | 18) BKD | 10) BKD | 15) BKD | 24) BKD |
| 12) GA-type 1 | 19) GA-type 1 | 11) GA-type 1 | 16) GA-type 1 | 25) GA-type 1 |
| 13) HMG | 20) HMG | | 17) HMG | 26) HMG |
| 14) IBD | 21) IBD | | 18) IBD | 27) IBD |
| 15) IVA | 22) IVA | 12) IVA | 19) IVA | IVA |
| • Acute onset | • Acute onset | | • Acute onset | 28) Acute onset |
| • Chronic onset | • Chronic onset | | • Chronic onset | 29) Chronic onset |
| 16) Malonic aciduria | 23) Malonic aciduria | | 20) Malonic aciduria | 30) Malonic aciduria |
| 17) MCD | 24) MCD | 13) MCD | | 31) MCD |
| 18) MHBD | 25) MHBD | | | |
| 19) MMA | MMA | 14) MMA | 21) MMA | MMA |
| • MMA, mut- | 26) MMA, mut- | | • (unspecified types) | 32) MMA, mut- |
| • MMA, mut 0 | 27) MMA, mut 0 | | | 33) MMA, mut 0 |
| • MMA, cbl A,B | 28) MMA, cbl A,B | | | 34) Some cbl types |
| • MMA, cbl C,D | 29) MMA, cbl C,D | | | 35) Maternal B12 deficiency |
| • MMA, cbl E | | | | |
| • MMA, cbl F | | | | |
| • MMA, cbl G | | | | |
| 20) PA | 30) PA | 15) PA | 22) PA | PA |
| • Acute onset | • Acute onset | | • Acute onset | 36) Acute onset |
| • Late onset | • Late onset | | • Late onset | 37) Late onset |
| **FAOD:** | **FAOD:** | **FAOD:** | **FAOD:** | **FAOD:** |
| 21) Carnitine uptake/transport defects[5] | 31) CAT | | 23) CAT | 38) CAT |
| | 32) CPT-type 1 | | | 39) CPT-type 1 |
| | 33) CPT-type 2 | | 24) CPT-type 2 | 40) CPT-type 2 |
| | 34) CUD | 16) CUD | | 41) CUD[3] |
| | | | | 42) DECRD[3] |
| 22) LCHADD | 35) LCHADD | 17) LCHADD | 25) LCHADD | 43) LCHADD |
| • TFP | 36) TFP | 18) TFP | 26) TFP | 44) TFP |
| 23) MADD | 37) MADD | | 27) MADD | 45) MADD |
| 24) MCADD | 38) MCADD | 19) MCADD | 28) MCADD | 46) MCADD |
| 25) SCADD | 39) SCADD | | | 47) SCADD |
| | | | 29) SCHADD | 48) SCHADD |
| 26) VLCADD | 40) VLCADD | 20) VLCADD | 30) VLCADD | 49) VLCADD |
| **Others:** | **Others:** | **Others:** | **Others:** | **Others:** |
| 27) BIO | | 21) BIO | | BIO |
| | | | | 50) Complete |
| | | | | 51) Partial |
| 28) CAH | CAH | 22) CAH | | 52) CAH |
| • Salt wasting | 41) Salt wasting | | | |
| • Virilizing | 42) Virilizing | | | |
| 29) CH | 43) CH | 23) CH | | |
| | 44) Variant CH | | | |
| | | 24) CF | | 53) CF |
| | 45) EE | | | |
| | | 25) G6PD | | 54) GP6PD |
| 30) GALT | 46) GALT | 26) GALT | | |
| | 47) Gyrate atrophy | | | |
| 31) HB | 48) HB (23 types) | | | |
| | 49) PRO | | | |
| 32) SCD | 50) SCD | 27) SCD | | |

## Amino Acid Disorder (AAD):

| | |
|---|---|
| ARG | Arginase deficiency |
| ASAL | Argininosuccinate lyase deficiency |
| ASAS | Argininosuccinate synthetase deficiency (aka: citrullinemia) |
| BPT | Biopterin cofactor deficiencies |
| CIT | Citrullinemia |
| CPS | Carbamoylphosphate synthetase deficiency |
| HCY | Homocystinuria (aka: cystathionine synthase deficiency) |
| HHH | Hyperammonemia,hyperornithinemia, homocitrullinuria syndrome |
| HOGA | Hyperornithinemia with gyral atrophy |
| HPHE | Hyperphenylalaninemia |
| MAT | Methionine adenosyltransferase deficiency (aka: hypermethioninemia) |
| MSUD | Maple syrup urine disease |
| NKH | Nonketotic hyperglycinemia |
| PKU | Phenylketonuria |
| TNTYR | Transient neonatal tyrosinemia |
| TYR | Tyrosinemia |

## Organic Acid Disorder (OAD):

| | |
|---|---|
| 2MBC | 2-methylbutyryl CoA dehydrogenase deficiency |
| 3MCC | 3-methylcrotonyl CoA carboxylase deficiency |
| 3MGH | 3-methylglutaconyl CoA hydratase deficiency (aka: 3-methylglutaconic aciduria) |
| BKD | Beta-ketothiolase deficiency (aka: mitochondrial acetoacetyl-CoA thiolase deficiency) |
| GA | Glutaric acidemia |
| HMG | 3-hydroxy-3-methylglutaryl CoA lyase deficiency |
| IBD | Isobutyryl CoA dehydrogenase deficiency |
| IVA | Isovaleric acidemia |
| MCD | Multiple carboxylase deficiency |
| MHBD | 2-methyl-3-hydroxybutyryl CoA dehydrogenase deficiency |
| MMA | Methylmalonic acidemia |
| PA | Propionic acidemia |

## Fatty Acid Oxidation Disorder (OAD):

| | |
|---|---|
| CAT | Carnitine/acylcarnitine translocase deficiency |
| CPT | Carnitine palmitoyl transferase deficiency |
| CUD | Carnitine uptake defect (aka: carnitine deficiency; carnitine transporter deficiency) |
| DECRD | 2,4-dienoyl-CoA reductase deficiency |
| LCHADD | Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency |
| MADD | Multiple acyl-CoA dehydrogenase deficiency (aka: glutaric acidemia, type 2) |
| MCADD | Medium chain acyl-CoA dehydrogenase deficiency |
| SCADD | Short chain acyl-CoA dehydrogenase deficiency |
| SCHADD | Short chain 3-hydroxyacyl-CoA dehydrogenase deficiency |
| TFP | Trifunctional protein deficiency |
| VLCADD | Very long chain acyl-CoA dehydrogenase deficiency |

## Other:

| | |
|---|---|
| BIO | Biotinidase deficiency |
| CAH | Congenital adrenal hyperplasia |
| CF | Cystic fibrosis |
| CH | Congenital hypothyroidism |
| EE | Ethylmalonic encephalopathy |
| G6PD | Glucose-6-phosphate dehydrogenase deficiency |
| GALT | Galactosemia |
| HB | Other hemoglobinopathies |
| PRO | Prolinemia |
| SCD | Sickle cell disease |

[1] as of mid-2005. Not all conditions are screened for, but all may be detected via screening.

[2] based on the StepOne™ supplemental screening program.

[3] the probability of detecting this in newborns is low.

[4] BPT screening is done on infants with a high phenylalanine level.

[5] is comprised of CAT, CPT-type 1, CPT-type 2, and CUD.