Newborn Screening Laboratory Manual of Services

Test Panel: Please see the following links for a detailed description of testing in the Newborn Screening section. Information about the Newborn Screening program is available here.

Endocrine Disorders
- Congenital adrenal hyperplasia (CAH)
- Congenital hypothyroidism (TSH)

Hemoglobinopathies
- Sickle cell disease (FS)
- Alpha (Barts)
- Sickle βeta Thalassemia (FSA)
- Other sickling hemoglobinopathies such as: FAS, FAC, FAD, FAE
- Homozygous conditions such as: FC, FD, FE

Metabolic Disorders
- Biotinidase deficiency
- Galactosemia
- Cystic fibrosis (CF) first tier screening for elevated immunoreactive trypsinogen (IRT)

Amino acid disorders
- Phenylketonuria (PKU) / Hyperphenylalaninemia
- Maple syrup urine disease (MSUD)
- Tyrosinemia, type 1 and possibly type 2 or type 3 - tyrosine levels may not be sufficiently elevated for detection
- Homocystinuria / Hypermethioninemia

Urea cycle disorders
- Citrullinemia (argininosuccinate synthetase deficiency)
- Argininosuccinic aciduria (argininosuccinate lyase deficiency)
- Argininemia

Organic acid disorders
- 2-methylbutyryl-CoA dehydrogenase deficiency (2MBD)
3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
3-hydroxy-3-methylglutaryl-CoA lyase deficiency (3HMG)
3-methylglutaconic aciduria (3MGA)
Glutaric aciduria, type 1 (GA1)
Propionic acidemia (PA)
Isovaleric acidemia (IVA)
Methylmalonic acidemia (MMA)
Malonic aciduria (MA)
Beta-ketothiolase deficiency (BKT)
Multiple carboxylase deficiency (MCD)

**Fatty acid oxidation disorders**
- Short chain acyl-CoA dehydrogenase deficiency (SCAD)
- Medium/Short chain L-3-hydroxyacyl-CoA-dehydrogenase deficiency (M/SCHAD)
- Isobutyryl-CoA dehydrogenase deficiency (IBCD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
- Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Trifunctional protein deficiency (TFPD)
- Carnitine palmitoyl transferase deficiency type 2 (CPT2)
- Carnitine palmitoyl transferase deficiency type 1 (CPT1A)
- Carnitine/acylcarnitine translocase deficiency (CACT) - neonatal form, extremely rare
- Carnitine uptake defect (CUD) - may not be reliably detected in first days of life
- Multiple acyl-CoA dehydrogenase deficiency (MADD) / Glutaric aciduria, type 2 (GA2)

**Severe Combined Immunodeficiency (SCID) and other T-Cell Lymphopenias** (screening for nascent T-cells; disorders detected can be, but are not limited to:
- All types of SCID
- Variant SCID
- Leaky SCID
- DiGeorge Syndrome
- Secondary T-cell lymphopenia
- CHARGE Syndrome
- Idiopathic T-cell lymphopenia
- Indicators of cardiac defects or cardiac surgery

**Lysosomal Storage Diseases**
- Fabry Disease
- Gaucher Disease
- Pompe Disease
- Krabbe Disease*
- Niemann Pick Disease
- Hurler's Disease (MPS-I)
- Hunter Syndrome (MPS-II)*
  *Hunter Syndrome and Krabbe Disease screening are currently being developed and will be added to the panel as soon as Laboratory Information support allows.

**Unsatisfactory Specimens** Unsatisfactory specimen reports indicate the specimen was
improperly collected, handled or submitted, as determined by the Department’s Division of Laboratories. Specimens must be of good quality to assure reliable, valid newborn screening; unsatisfactory specimens require collection and submission of a new sample to assure that every baby receives a valid newborn screening. Unsatisfactory results are reported from the program by a letter indicating the nature of the specimen and the need for immediate repeat specimen collection. The letter is sent by mail to the submitting physician or facility. Additional information about specimen collection and submission is available here.

**Turn Around Time:** 4 days for abnormal test results and 10 days for normal test. Positive or abnormal results are provided as quickly as possible.

**Ship to:** Chicago IDPH Lab, 2121 West Taylor, Chicago, IL 60612

**Shipping Kits:** Call the Springfield Laboratory at 217-782-6562. Call for more information about this service 217-785-8101.

**Submission Form:** Newborn Screening Submission Form