



Cook Children's Genetics Center

Metabolic Genetics Services

By Alice Basinger, M.D., Ph.D.

Why refer for an abnormal newborn screening result?

Metabolic disorders are rare, and this makes them hard to recognize clinically. A few drops of blood on a paper card provide valuable information on treatable metabolic disorders for our infants. Texas newborn screening began with testing for phenylketonuria (PKU) in 1965. As testing techniques were optimized, more disorders were added. In 2007, the Texas Department of State Health Services expanded newborn screening to include 29 disorders. In 2010, testing for Cystic Fibrosis was added and in 2012, testing for severe combined immunodeficiency was added.

In 2011, Texas newborn screening helped diagnose a metabolic disorder in one of every 5,000 infants tested. The majority of these disorders do not have visible signs at birth or in early life. Early treatment can limit or reduce the effects of the metabolic disorder on growth and development.



Here is a list of the primary metabolic disorders detectable on Texas newborn screening and handled by Cook Children's Metabolic Genetics Services:

- **Amino acid disorders**

- Argininosuccinic aciduria
- Citrullinemia
- Homocystinuria
- Maple syrup urine disease (MSUD)
- Phenylketonuria (PKU)
- Tyrosinemia type 1

- **Biotinidase deficiency**

- **Fatty acid oxidation disorders**

- Carnitine palmitoyl transferase 1 (CPT1) deficiency
- Carnitine uptake disorder
- Long-chain hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
- Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
- Trifunctional protein (TFP) deficiency
- Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency

- **Galactosemia**

- **Organic acid disorders**

- Beta-ketothiolase deficiency
- Glutaric acidemia type 1
- Isovaleric acidemia
- Methylmalonic acidemia
- Multiple carboxylase deficiency
- Propionic acidemia
- 3-OH-3-methylglutaric aciduria
- 3-methylcrotonylcarboxylase deficiency



These disorders found by the newborn screening are handled by other Cook Children's specialty services:

- Congenital adrenal hyperplasia and hypothyroidism (refer to Endocrinology team)
- Cystic Fibrosis (refer to Pulmonary team)
- Hemoglobinopathies (refer to Hematology team)
- Severe combined immunodeficiency (refer to Infectious Disease team)

Confirmatory testing:

- An abnormal screen is not diagnostic—further testing is needed to confirm the diagnosis
- Confirmatory testing follows a recommended pattern for each type of abnormal screening
- Tests may evaluate metabolites in blood and urine and/or enzyme activities in blood
- Most confirmatory testing is done through referral labs and has a 1-3 week turnaround time

Why refer for an abnormal metabolic screening result:

- Initial education with family done with metabolic genetics team (includes two metabolic geneticists, metabolic dietitian and metabolic nurse)
- Less time needed by primary care physician for family education on metabolic disorders
- Labs ordered by and returned to metabolic team for efficient flow and diagnosis
- Medication and/or special diet can be started as needed
- Special circumstances, such as maternal metabolic disease altering the child's screening results, are addressed by the metabolic team
- Ease of follow-up and familiarity with the metabolic team for families whose children are diagnosed with metabolic disorders

A referral for an abnormal newborn screening should answer the following questions:

- What does this abnormal screening mean?
- What further testing is needed?
- What is needed to protect the child while testing is in progress?
- What treatments may be needed later?
- What health effects are expected for a child diagnosed with this disorder?
- What is the risk of this disorder occurring in other children in the family?

How to refer for abnormal metabolic screens

- The team at Cook Children's Metabolic Genetics Center can see children in our Fort Worth clinic or by telemedicine in Abilene, Denton, Midland, San Angelo and Waco
- Call 682-885-2170 to schedule an appointment
- Call 1-800-KID-HURT for transport or phone consult



Metabolic geneticists:

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