

SCREENING TESTS

SCREENING TESTS PERFORMED IN KANSAS

There are a number of disorders for which cost effective newborn screening is feasible. Each state decides which disorders it wishes to screen. While it is possible to screen for over 150 different disorders using a few drops of blood applied to special filter paper, it is not now practical to screen for all of these disorders. Currently in Kansas, the Newborn Screening Program screens for the twenty-nine of the thirty core panel of hereditary disorders recommended by the American College of Medical Genetics (ACMG). These disorders include:

Amino Acid Disorders

- Phenylketonuria (PKU)
- Maple Syrup Urine Disease (MSUD)
- Homocystinuria (HCY)
- Tyrosinemia Type I (TYR I)
- Argininosuccinic acidemia (ASA)
- Citrullinemia (CIT)

Fatty Acid Disorders

- Medium chain Acyl-CoA Dehydrogenase deficiency (MCAD)
- Very Long chain Acyl-CoA Dehydrogenase deficiency (VLCAD)
- Long Chain Hydroxy Acyl-CoA Dehydrogenase deficiency (LCHAD)
- Trifunctional protein deficiency (TFP)
- Carnitine uptake defect (CUD)

Organic Acid Disorders

- Isovaleric Acidemia (IVA)
- Glutaric Aciduria Type I (GA-I)
- 3-hydroxy-3-methylglutaryl CoA lyase deficiency (HMG)
- Multiple carboxylase deficiency (MCD)
- Methylmalonic Acidemia/Methylmalonyl-CoA mutase (MUT)
- Methylmalonic Acidemia/Vitamin B12 Disorders (Cbl A,B)
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
- Propionic Acidemia (PROP)
- Beta ketothiolase deficiency (BKT)

Hemoglobinopathies

- Sickle Cell Anemia (SCA)
- Sickle C Disease (HB S/C)
- Sickle Beta Thalassemia (HB S/Th)

Other Disorders

- Congenital Hypothyroidism (CH)
- Biotinidase deficiency (BIO)
- Congenital Adrenal Hyperplasia (CAH)
- Transferase Deficient Galactosemia (GALT)
- Cystic Fibrosis (CF)
- Hearing (HEAR)

Currently, Kansas does not screen for Severe Combined Immune Deficiency (SCID) which was added to the core panel in May, 2010.

Practitioners are urged to use the term "Newborn Screening" rather than "PKU test" since other disorders besides PKU are included in the screening battery. Not all screened disorders are metabolic.

TABLE 1
Summary of Disorders Screened by the Program

Condition	Test Performed	Birth Incidence Rate	Symptoms if Not Treated	Treatment
Phenylketonuria (PKU)	MS/MS test for Phenylalanine	> 1 in 25,000	Severe mental retardation, seizures.	Low phenylalanine diet
Maple Syrup Urine Disease (MSUD)	MS/MS test for Leucine	< 1 in 100,000	Seizures, coma and death if not treated. Urine smells like maple syrup.	Restricted protein diet; special supplements
Homocystinuria (HCY)	MS/MS test for Methionine	< 1 in 100,000	Mental retardation, eye problems, skeletal abnormalities and stroke.	Special diet, vitamins B6 or B12 and betaine
Tyrosinemia (TYR I)	MS/MS test for Tyrosine	< 1 in 100,000	Build-up of succinylacetone in the liver. Liver or kidney failure, nerve damage and death.	Drug treatment, sometimes with a low-protein diet, can prevent liver and kidney damage in most patients.
Argininosuccinic Acidemia (ASA)	MS/MS test for Argininosuccinic Acid	< 1 in 100,000	Build-up of argininosuccinic acid and ammonia resulting in brain swelling, coma and sometimes death.	Low protein diet, avoid fasting, medications to prevent ammonia build-up, nutritional supplements and sometimes liver transplant.
Citrullinemia (CIT)	MS/MS test for Citrulline	< 1 in 100,000	Build-up of citrulline and ammonia during newborn period and infancy can result in seizures, coma, brain damage and death.	Low protein diet, medication to prevent ammonia build-up and nutritional supplements.
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)	MS/MS test for C8; C6,C10	> 1 in 25,000	Infants seem well, then suddenly develop seizures due to low blood sugar; liver failure, coma and death.	Avoidance of fasting and addition of nutritional supplements.

Condition	Test Performed	Birth Incidence Rate	Symptoms if Not Treated	Treatment
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)	MS/MS test for C14:1	> 1 in 75,000	Infants develop heart and liver failure and can die during the first year of life.	High carbohydrate/low fat diet, nutritional supplements, and the avoidance of fasting and prolonged exercise.
Long-chain 3-OH acyl-CoA dehydrogenase deficiency (LCHAD)	MS/MS test for C16-OH +/- C18:1-OH	> 1 in 75,000	Symptoms resulting in heart, lung, or liver failure and death can occur soon after birth.	High carbohydrate/low fat diet, nutritional supplements and avoidance of fasting.
Trifunctional protein deficiency (TFP)	MS/MS test for C16-OH +/- C18:1-OH	< 1 in 100,000	Seemingly well infant can die suddenly of apparent SIDS or develop low muscle tone, seizures, heart failure, coma, often following illness.	Strict avoidance of fasting, a low fat diet and nutritional supplements.
Carnitine uptake defect (CUD)	MS/MS test for C0	< 1 in 100,000	Symptoms include hypoglycemia and sudden unexpected death in infancy.	Early diagnosis and treatment with carnitine permits normal development.
Isovaleric Acidemia (IVA)	MS/MS test for C5	< 1 in 100,000	Coma, permanent brain damage and death	Low protein diet and nutritional supplements
Glutaric Acidemia Type I (GA-I)	MS/MS test for C5-DC	> 1 in 75,000	A trigger such as a viral disease leads to brain damage, seizures, low muscle tone and death.	Dietary protein restriction and L-carnitine supplement along with the prompt treatment of illness/fever can prevent brain damage
HMG-CoA lyase deficiency (HMG)	MS/MS test for C5-OH	< 1 in 100,000	Low blood sugar and accumulation of organic acids can result in brain damage, mental retardation, coma and death	Avoid fasting; a diet low in leucine and fat and high in carbohydrates can lead to normal development.

Condition	Test Performed	Birth Incidence Rate	Symptoms if Not Treated	Treatment
Multiple Carboxylase Deficiency (MCD)	MS/MS test for C5-OH	< 1 in 100,000	Build up of lactic acid and other organic acids. Symptoms include skin rashes, hair loss and brain damage, coma and death without treatment.	Biotin supplements can result in normal growth and development.
Methylmalonic Acidemia – Mutase Deficiency (MUT)	MS/MS test for C3	> 1 in 75,000	Illness in the first week of life, death in the first month of life.	Special metabolic formula/low protein diet and medication minimize but cannot prevent central nervous system dysfunction.
Methylmalonic Acidemias (Cbl A & B)	MS/MS test for C3	< 1 in 100,000	Build-up of organic acids in the blood results in brain damage, seizures, paralysis, coma and death. Symptoms may occur in first week, but most affected individuals remain symptom-free	Treatment of B12 injections and a special metabolic formula/low-protein diet can prevent serious problems.
3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)	MS/MS test for C5-OH	> 1 in 75,000	Inability to process leucine can lead to brain damage, seizures, liver failure and death in infancy or no symptoms at all into adulthood.	Treatment with a low-protein diet; in some cases, nutritional supplements may be helpful.
Propionic Acidemia (PROP)	MS/MS test for C3	> 1 in 75,000	Brain damage, coma and death.	Low-protein diet and nutritional supplements.
Beta-Ketothiolase Deficiency (BKT)	MS/MS test for C5-OH	< 1 in 100,000	Episodes of organic acid build-up, often triggered by childhood illness can progress to coma, brain damage and death.	Prompt IV treatment to keep blood sugar levels up and blood acid levels down during illness, children can develop normally.

Condition	Test Performed	Birth Incidence Rate	Symptoms if Not Treated	Treatment
Sickle Cell Anemia	Hemoglobin electrophoresis	> 1 in 5,000; 1 in 400 in African Americans	Death or disability from various complications of sickle cell disease and related disorders	Antibiotic prophylaxis; Early family education; Early treatment of complications
Hb S/ Beta Thalassemia	Hemoglobin electrophoresis	> 1 in 50,000	Form of sickle cell anemia. Symptoms are often milder than Hb SS, but severity varies.	Routine treatment with penicillin may not be universally recommended.
Hb S/C disease	Hemoglobin electrophoresis	> 1 in 25,000	Form of sickle cell anemia, often milder than Hb SS.	Routine treatment with penicillin may not be universally recommended.
Congenital Hypothyroidism (CH)	TSH assay	> 1 in 5,000	Mental retardation, other brain damage, growth delay	Thyroid hormone
Biotinidase Deficiency (BIOT)	Colorimetric test of enzyme activity	> 1 in 75,000	Frequent infections, skin rash, uncoordinated movement, hearing loss, seizures and mental retardation. Left untreated, the deficiency can lead to coma and death.	Oral doses of biotin
Congenital Adrenal Hyperplasia (CAH)	17-OHP	> 1 in 25,000	Salt wasting forms can be life-threatening due to loss of salt.	Salt and hormone replacement
Galactosemia (GALT)	Modified Beutler test for galactose-1-phosphate uridyl transferase enzyme	> 1 in 50,000	Galactose accumulates and damages vital organs, leading to blindness, severe mental retardation, infection and death.	Milk and dairy products must be eliminated from diet for life. Even with treatment, a chance for developmental delays exists.

Condition	Test Performed	Birth Incidence Rate	Symptoms if Not Treated	Treatment
Cystic Fibrosis (CF)	Immunoreactive trypsinogen (IRT)/DNA PCR	> 1:5,000	Lung and digestive problems, often beginning in early infancy. Recurrent pneumonia and failure to thrive are common.	Early diagnosis and treatment may improve the child's growth. Treatment depends on severity – includes high-calorie diet supplemented with vitamins and medication to improve digestion; respiratory therapy.