

Georgia's Newborn Screening Panel (Disorders)

Organic Acid	Fatty Acid Oxidation	Amino Acid	Hemoglobinopathy	Other
<ul style="list-style-type: none"> • Beta Ketothiolase (BKT) • Cobalamin A and B Deficiency (Cbl A,B) • Glutaric Acidemia type I • 3OH 3-CH3 Glutaric Aciduria (HMG) • Isovaleric Acidemia (IVA) • 3 Methylcrotonyl-Co A Carboxylase Deficiency (3MCC) • Multiple Carboxylase Deficiency • Methylmalonic Acidemia (MMA) • Propionic Acidemia 	<ul style="list-style-type: none"> • Carnitine Uptake Defect • Long Chain 3 hydroxyl acyl-CoA dehydrogenase Deficiency • Medium Chain acyl-CoA dehydrogenase Deficiency • Trifunctional Protein Deficiency • Very Long chain acyl-CoA dehydrogenase Deficiency 	<ul style="list-style-type: none"> • Argininosuccinic Acidemia • Citrullinemia • Homocystinuria • Maple Syrup Urine Disease • Phenylketonuria • Tyrosinemia 	<ul style="list-style-type: none"> • Sickle Cell Disease • Sickle SC Disease • Sickle Beta Thalassemia 	<ul style="list-style-type: none"> • Biotinidase • Congenital Adrenal Hyperplasia • Congenital Hypothyroidism • Cystic Fibrosis • Galactosemia

3-methylcrotonyl-CoA carboxylase deficiency (3-MCC)

(3-methel-crow-ton-eel co-A car-box-il-ace de-fish-in-sea)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen? Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called 3-MCC. There are many other conditions that can cause a similar positive result on newborn screening.

A positive newborn screen does not mean your baby has 3-MCC or any of the other conditions above, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like 3-MCC.

Sometimes, a baby has a positive newborn screen because the mother has a hidden form of 3-MCC. Mothers of babies with positive newborn screens also need testing.

What problems can 3-MCC cause?

3-MCC is different for each child. Some children with 3-MCC have fewer health problems, while other children may have very serious complications.

If 3-MCC is not treated, a child might develop:

Serious illness (metabolic crisis)

Muscle weakness

Sleepiness

Seizures

Poor growth

Mental retardation

Coma

It is very important to follow the doctor's instructions for testing and treatment. Many people with 3-MCC have no obvious problems so they don't even know they have it. This is why mothers of babies with 3-MCC need testing.

What is 3-MCC?

3-MCC affects a special enzyme needed to break down proteins from the food we eat so they can be used for energy and growth. In 3-MCC, the enzymes used to break down proteins are missing or not working properly. A person with 3-MCC doesn't have enough enzyme to break down protein containing leucine, which can cause harmful toxins to build up in the body.

3-MCC is a disorder that is passed on, or inherited, from a child's mother and father. Because 3-MCC is a genetic disease, family members are at risk of having 3-MCC too, even if no one in the family has had it before.

What is the treatment for 3-MCC?

3-MCC can be treated. The treatment is life-long. Treatment for children with 3-MCC can include:

Medications to help the body get rid of harmful toxins

Low-protein diet - a dietician will help you set up the best diet for your child

Children with 3-MCC should see their regular doctor, a doctor who specializes in 3-MCC, and a dietician. Children with 3-MCC can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>.

****This information is provided for the families and physicians of children newly identified with, or at an increased risk for, a metabolic condition as determined by the [Georgia Newborn Screening Program](#). The links provided are to information from recognized and trusted sources. It is however, important to note that this content is NOT a substitute for professional medical advice, diagnosis or treatment.**

Argininosuccinic Acidemia (ASA)

(Are-gin-in-o-suk-sin-ic acid-ee-me-ah)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called ASA.

A positive newborn screen does not mean your baby has ASA, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like ASA.

What problems can ASA cause?

ASA is different for each child. Some children have a mild form of ASA with fewer health problems, while other children may have a severe form of ASA with serious complications.

If severe ASA is not treated, a child might develop:

Feeding problems

Sleepiness

Vomiting

Muscle weakness

Seizures

Swelling of the brain

Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is ASA?

A person with ASA doesn't have enough enzyme to remove ammonia from the body. Ammonia is very harmful and can cause health problems if it isn't removed from the body every day.

ASA is a disorder that is passed on, or inherited, from a child's mother and father. Because ASA is a genetic disease, family members are at risk of having ASA too, even if no one in the family has had it before.

What is the treatment for ASA?

ASA can be treated. The treatment is life-long. Treatment for children with ASA can include:

Medications to help prevent high ammonia

Special formula low in protein

Low protein diet - a dietician will help you set up the best diet for your child

Children with ASA should see their regular doctor, a doctor who specializes in ASA, and a dietician.

Children with ASA can benefit from prompt and careful treatment.

Resources for parents

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Georgia Department of Public Health

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Beta-Ketothiolase Deficiency (BKT)

(Bay-tah- key-toe-thigh-o-lase de-fish-in-sea)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called BKT.

A positive newborn screen does not mean your baby has BKT, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like BKT.

What problems can BKT cause?

BKT is different for each child. Some children have a mild form of BKT with fewer health problems, while other children may have a severe form of BKT with serious complications and some children with BKT never develop symptoms.

If BKT is not treated, a child might develop:

Extreme sleepiness or lack of energy

Vomiting

Diarrhea

Fever

Poor appetite

Ketones in the urine (substances created during the breakdown of fat)

Low blood sugar

Increased levels of acidic substances in the blood

Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is BKT?

BKT affects a special enzyme needed to break down proteins from the food we eat so they can be used for energy and growth. In BKT, the enzyme used to break down proteins are missing or not working properly. A child with BKT doesn't have enough enzyme to break down proteins containing isoleucine, which can cause harmful toxins to build up in the body.

BKT is a disorder that is passed on, or inherited, from a child's mother and father. Because BKT is a genetic disease, family members are at risk of having BKT too, even if no one in the family has had it before.

What is the treatment for BKT?

BKT can be treated. The treatment is life-long. Treatment for children with BKT can include:

Medications to help prevent the body get rid of harmful toxins

Low protein diet - a dietician will help you set up the best diet for your child

Avoid going a long time without food

Children with BKT should see their regular doctor, a doctor who specializes in BKT, and a dietician.

Children with BKT can benefit from prompt and careful treatment.

Resources for parents

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Biotinidase Deficiency

(bye-o-tin-ah-daze de-fish-in-sea)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called biotinidase deficiency.

A positive newborn screen does not mean your baby has biotinidase deficiency, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing with specialists in disorders like biotinidase deficiency.

What is biotinidase deficiency?

Biotinidase deficiency affects an enzyme needed to free biotin (one of the B vitamins) from the food we eat, so it can be used for energy and growth.

A person with biotinidase deficiency doesn't have enough enzyme to free biotin from foods so it can be used by the body.

Biotinidase deficiency is a disorder that is passed on, or inherited, from a child's mother and father. Because biotinidase deficiency is

a genetic disease, family members are at risk of

having biotinidase deficiency too, even if no one in the family has had it before.

What problems can biotinidase deficiency cause?

Biotinidase deficiency is different for each child. Some children have a mild, partial biotinidase deficiency with few health problems, while other children may have complete biotinidase deficiency with serious complications.

If biotinidase deficiency is not treated, a child might develop:

Muscle weakness

Hearing loss

Vision (eye) problems

Hair loss

Skin rashes

Seizures

Developmental delay

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for biotinidase deficiency?

Biotinidase deficiency can be treated. The treatment is life-long. Treatment for children with biotinidase deficiency includes:

Daily biotin vitamin pill(s) or liquid

Children with biotinidase deficiency should see their regular doctor, and a doctor who specializes in biotinidase deficiency.

With prompt and careful treatment, children with biotinidase deficiency have a good chance to live healthy lives with normal growth and development.

Resources for parents

National Organization for Rare Disorders

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Emory Genetics Lab
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Carnitine Uptake/Transport Deficiency (CUD/CTD)

(car-na-teen up-take/tran-sport de-fish-in-sea)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have CUD/CTD (cee-you-dee/cee-tee-dee).

A positive newborn screen does not mean your baby has CUD/CTD, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like CUD/CTD. Sometimes, a baby has a positive screen because the mother has a hidden form of CUD/CTD. Mothers of babies with positive newborn screens also need testing.

What is CUD/CTD?

CUD/CTD affects an enzyme needed to break down fats in the food we eat, so they can be used for energy and growth. In CUD/CTD, an enzyme used to break down fats into energy is missing or not working properly. A person with CUD/CTD doesn't have enough enzyme to break down fat into carnitine. Carnitine helps the body make energy from fats and also helps the body use stored fat. CUD/CTD is a disorder that is passed on, or inherited, from a child's mother and father. Because CUD/CTD is a genetic disease, family members are at risk

of having CUD/CTD too, even if no one in the family has had it before.

What problems can CUD/CTD cause?

CUD/CTD is different for each child. Some children with CUD/CTD do not have any health problems, while other children may have very serious complications.

If CUD/CTD is not treated, a child might develop:

Sleepiness

Behavior changes (such as crying for no reason)

Feeding problems

Seizures

Coma

It is very important to follow the doctor's instructions for testing and treatment. Some people with CUD/CTD have no obvious problems, so they don't even know they have it. This is why mothers of babies with positive screens need testing.

What is the treatment for CUD/CTD?

CUD/CTD can be treated. The treatment is life-long. Treatment for children with CUD/ CTD can include:
Carnitine to help the body make energy and get rid of harmful toxins
Frequent meals/snacks - a dietician will help you set up the best diet for your child
Low fat/High carb diet - a dietician will help you learn what foods can be eaten

Children with CUD/CTD should see their regular doctor, a doctor who specializes in CUD/CTD, and a dietician.

Children with CUD/CTD can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

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National Urea Cycle Disorders Foundation

www.nucdf.org

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Citrullinemia (CIT) **(sit-roo-lin-ee-me-ah)**

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen shows an increase in a component of protein called citrulline. This result suggests your baby might have a disorder called citrullinemia. There is another form of citrullinemia that can also cause an increase in citrulline on newborn screening. This form is very rare and is called citrullinemia, type II.

A positive newborn screen does not mean your baby has citrullinemia, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like citrullinemia.

What is citrullinemia?

Citrullinemia affects an enzyme needed to break down certain proteins and remove waste ammonia from the body so it doesn't build-up and cause health problems.

A person who has citrullinemia doesn't have enough enzyme to break down protein containing citrulline or remove ammonia from the body. Both citrulline and ammonia are very harmful to the body and can cause health problems if they build up.

Citrullinemia is a disorder that is passed on, or inherited, from a child's mother and father. Because citrullinemia is a genetic disease, family members are at risk of having citrullinemia too, even if no one in the family has had it before.

What problems can citrullinemia cause?

Citrullinemia is different for each child. Some children have a mild form of citrullinemia with fewer health problems, while other children may have a severe form of citrullinemia with serious complications.

If citrullinemia is not treated, a child might develop:

- Feeding problems
- Sleepiness
- Vomiting
- Muscle weakness
- Seizures
- Swelling of the brain
- Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for citrullinemia?

Citrullinemia can be treated. The treatment is life-long. Treatment for children with citrullinemia can include:

- Medications to help prevent high ammonia
- Special formula low in protein
- Low protein diet - a dietician helps families set up the best diet for their child

Children with citrullinemia should see their regular doctor, a doctor who specializes in citrullinemia, and a dietician.

Children with citrullinemia can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

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Georgia Department of Public Health

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Congenital Adrenal Hyperplasia (CAH) **(kuhn-jen-i-til uh-dreen-l hi-purr-play-she-uh)**

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called CAH.

A positive newborn screen does not mean your baby has CAH, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing for CAH.

What is CAH?

CAH affects the way the body makes chemicals called hormones. Hormones help the body work and grow properly.

A person with CAH doesn't make enough of a hormone called cortisol (also known as the "stress hormone"). Without enough cortisol, the body can have problems growing and developing. Some children may also have trouble keeping the right amount of salt in the body. The wrong amount of salt can cause the body to stop working properly.

CAH is a disorder that is passed on, or inherited, from a child's mother and father. Because CAH is a genetic disease, family members are at risk of having CAH too, even if no one in the family has had it before.

What problems can CAH cause?

CAH is different for each child and affects females and males in different ways. Girls with CAH can have problems with their genitals, while boys with CAH don't. There are three main types of CAH:

Salt-wasting CAH

Simple virulizing CAH

Nonclassic CAH

If **salt-wasting CAH** is not treated, a child might develop:

Poor feeding and weight gain

Sleepiness

Dehydration (not enough fluid)

Life-threatening salt-wasting crises

If **simple virulizing CAH** is not treated, a child might develop:

Rapid growth in childhood

Early puberty

Individuals with **nonclassic CAH** have milder problems that don't start until childhood, or even adulthood. It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for CAH?

CAH can be treated. The treatment is life-long. Treatment for children with CAH includes:

Daily medications to replace missing hormones

Children with CAH should see their regular doctor and a pediatric endocrinologist (hormone specialist). With prompt and careful treatment, children with CAH can live a healthy, normal life.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Cystic Fibrosis (CF) **(sis-tick fye-bro-sis)**

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen shows your baby has a chance of having cystic fibrosis (CF).

Most likely, a positive newborn screen does not mean your baby has CF, but your baby needs more testing to know for sure.

Your baby's doctor will help arrange for special testing (called a sweat test) with a CF specialist when your baby is about 1 month old.

What is cystic fibrosis?

CF affects breathing and digestion (breaking down food).

A person with CF makes thick, sticky mucus that blocks the airways of the lungs, making it hard to breathe. This mucus can also make it harder for the body to break down food. In people with CF, the sweat glands also make very salty sweat.

CF is a disorder that is passed on or inherited from a child's mother and father. Because CF is a genetic disease, family members are at risk of having CF too, even if no one in the family has had it before.

What problems can cystic fibrosis cause?

CF is different for each child. Some children with CF have fewer health problems, while other children may have serious complications.

Children with CF develop:

- Coughing and wheezing
- Lung infections
- Poor growth and weight gain
- Greasy or oily stools

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for cystic fibrosis?

Although CF cannot be cured, many things can be done to treat the symptoms of CF. The treatment is life-long. Treatment for children with CF includes:

- Healthy, high-calorie diet
- Vitamins
- Medications to help absorb more nutrients from food
- Medications to prevent infection and help with breathing
- Help clearing mucus from the lungs

Children with CF should see their regular doctor and a doctor who specializes in CF.

Resources for parents

National Organization for Rare Disorders
www.rarediseases.org

National Urea Cycle Disorders Foundation
www.nucdf.org

Emory Genetics Lab
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Georgia Department of Public Health
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Galactosemia **(ga-lac-toe-see-me-ah)**

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called galactosemia. There are other forms of galactosemia that can also cause a positive result on newborn screening. These forms are very rare and are called Galactose Epimerase Deficiency and Galactokinase Deficiency.

A positive newborn screen does not mean your baby has galactosemia, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing with specialists in disorders like galactosemia.

What problems can galactosemia cause?

Galactosemia is different for each child. Some children have mild galactosemia with only a few health problems, while other children may have serious complications.

If galactosemia is not treated, a child might develop:

- Serious infection
- Liver failure
- Mental retardation
- Poor growth
- Cataracts (cloudiness) in the eyes

Children and adults treated for galactosemia may still have problems with:

- Speech and language
- Vision (eyesight)
- Growth
- Learning

It is very important to follow the doctor's instructions for testing and treatment.

What is galactosemia?

Galactosemia affects an enzyme needed to break down a sugar that is part of all milk products, including breast milk and most formulas. Galactosemia is a much more serious problem than lactose intolerance.

A person who has galactosemia doesn't have enough enzyme to break down the sugar known as galactose. If galactose is not broken down, it builds up in the body and causes problems. Galactosemia is a disorder that is passed on, or inherited, from a child's mother and father. Because galactosemia is a genetic disease, family members are at risk of having galactosemia too, even if no one in the family has had it before.

What is the treatment for galactosemia?

Galactosemia can be treated. The treatment is life-long. Treatment for children with galactosemia includes: Restriction of milk or foods that have milk in them - a dietician will help you set up the best diet for your child. Children with galactosemia should see their regular doctor, a doctor who specializes in galactosemia, and a dietician.

Children with galactosemia can benefit from prompt and careful treatment

Resources for parents

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Glutaric Acidemia, type 1 (GA1) **(glue-ter-ic acid-ee-me-ah)**

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called GA1 (gee-A-1).

A positive newborn screen does not mean your baby has GA1, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like GA1.

What is glutaric acidemia?

GA1 affects an enzyme needed to break down proteins from the food we eat, so they can be used for energy and growth. In GA1, the enzyme used to break down proteins is missing or not working properly.

A person with GA1 doesn't have enough enzyme to break down protein containing lysine and tryptophan, which causes a substance called glutaric acid to build up in the body. Glutaric acid is harmful to the body and causes health problems if it builds up.

GA1 is a disorder that is passed on, or inherited, from a child's mother and father. Because GA1 is a genetic disease, family members are at risk of having GA1 too, even if no one in the family has had it before.

What problems can glutaric acidemia cause?

GA1 is different for each child. Some children with GA1 have only a few health problems, while other children may have very serious complications. Babies with GA1 are usually healthy at birth, but many are born with a large head.

If GA1 is not treated, a child might develop:

Sleepiness

Feeding problems

Vomiting

Muscle weakness

Bleeding into the brain or eyes

Cerebral palsy

Seizures

Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for glutaric acidemia?

GA1 can be treated. The treatment is life-long. Treatment for children with GA1 can include:

Medications to help the body use protein and remove glutaric acid

Medications to help the body use energy

Diet low in lysine and tryptophan - a dietician will help you set up the best diet for your child

Children with GA1 should see their regular doctor, a doctor who specializes in GA1, and a dietician.

Children with GA1 can benefit from prompt and careful treatment.

Resources for parents

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3-OH-3-CH₃ Glutaric Aciduria,(HMG) (glue-tar-ic acid-ore-ia)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called HMG.

A positive newborn screen does not mean your baby has HMG, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like HMG.

What problems can HMG cause?

HMG is different for each child. Some children have a mild form of HMG with fewer health problems, while other children may have a severe form of HMG with serious complications and some children with HMG never develop symptoms.

If HMG is not treated, a child might develop:

Extreme sleepiness or lack of energy

Vomiting

Diarrhea

Fever

Poor appetite

Muscle weakness

Seizures

High levels of ammonia in the blood

Low blood sugar

Increased levels of acidic substances in the blood

Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is HMG?

HMG affects a special enzyme needed to break down proteins from the food we eat so they can be used for energy and growth. In HMG, the enzyme used to break down proteins are missing or not working properly. A child with HMG doesn't have enough enzyme to break down proteins containing leucine, which can cause harmful toxins to build up in the body.

HMG is a disorder that is passed on, or inherited, from a child's mother and father. Because HMG is a genetic disease, family members are at risk of having HMG too, even if no one in the family has had it before.

What is the treatment for HMG?

HMG can be treated. The treatment is life-long. Treatment for children with HMG can include:

Medications to help the body get rid of harmful toxins

Low leucine diet - a dietician will help you set up the best diet for your child

Avoid going a long time without food

Children with HMG should see their regular doctor, a doctor who specializes in HMG, and a dietician.

Children with HMG can benefit from prompt and careful treatment.

Resources for parents

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Sickle Cell Disease

(sik-el sel di-zez)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn

screen suggests your baby likely has a disorder called sickle cell disease.

A positive newborn screen means your baby likely has sickle cell disease and needs more testing to know for sure.

Your baby's doctor will help arrange for more testing with specialists in sickle cell disease.

What is sickle cell disease?

Sickle cell disease is a disorder that is passed on, or inherited, from a child's mother and father. Because sickle cell disease is a genetic disease, family members are at risk of having sickle cell disease too, even if no one in the family has had it before.

Sickle cell disease can be treated. Treatments for children with sickle cell disease can include:

Antibiotics to prevent infection

Medications for pain

Immunizations

Children with sickle cell disease should see their regular doctor and a doctor who specializes in sickle cell disease.

Children with sickle cell disease can benefit from prompt and careful treatment.

What problems can sickle cell disease cause?

Sickle cell disease is different for each child. Some children with sickle cell disease have only a few health problems, while other children may have serious complications.

If sickle cell disease is not treated, a child might develop:

Infections

Stroke

Drop in red blood cell count (anemia)

Painful crises

Enlarged spleen

It is important to follow the doctor's instructions for testing and treatment.

What is the treatment for sickle cell disease?

Sickle cell disease affects the red blood cells so they can't carry as much oxygen to the body. In sickle cell disease, the red blood cells are sickle (banana) shaped instead of round (donut) shaped. The sickled red blood cells can get trapped in the blood vessels and block blood flow. This can be painful and cause health problems.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>

Homocystinuria

(ho-mo-sis-te-en)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby likely has a disorder called Homocystinuria.

A positive newborn screen means your baby likely has Homocystinuria and needs more testing to know for sure.

Your baby's doctor will help arrange for more testing with specialists in Homocystinuria.

What is Homocystinuria?

Homocystinuria is one type of [amino acid disorder](#). People with this condition have problems breaking down an [amino acid](#) called [methionine](#) from the food they eat. Amino acid disorders (AAs) are a group of rare inherited conditions. They are caused by [enzymes](#) that do not work properly. Protein is made up of smaller building blocks called [amino acids](#). A number of different enzymes are needed to process these amino acids for use by the body. Because of missing or non-working enzymes, people with amino acid disorders cannot process certain amino acids. These amino acids, along with other toxic substances, then build up in the body and cause problems.

What problems can Homocystinuria cause?

Homocystinuria disease is different for each child. Some children with Homocystinuria have only a few health problems, while other children may have serious complications.

If Homocystinuria is not treated, a child might develop:

- poor growth
- problems gaining weight
- delays in crawling, walking, and talking
- behavior and emotional problems
- serious learning disabilities or mental retardation

It is important to follow the doctor's instructions for testing and treatment.

What is the treatment for Homocystinuria?

Homocystinuria can be treated. The treatment is life-long. Treatment for children with Homocystinuria can include:

- Low-methionine diet
- Medical Foods and Formula
- Supplements such as Vitamin B6, Betaine, Vitamin B12, Folic Acid, and L- Cystine Vitamin B6
- Frequent urine and blood test

Children with Homocystinuria should see their regular doctor, a doctor who specializes in Homocystinuria, and a dietician.

Children with Homocystinuria can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>

Hypothyroidism

(hi-po-thi-roi-diz-em)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby likely has a disorder called Hypothyroidism.

A positive newborn screen means your baby likely has Hypothyroidism and needs more testing to know for sure.

Your baby's doctor will help arrange for more testing with specialists in Hypothyroidism.

What is Hypothyroidism?

Hypothyroidism is a condition in which the person does not make enough thyroid [hormone](#). The thyroid gland is a butterfly-shaped organ at the base of the neck. Its job is to make specific hormones that help the cells of the body function correctly. The main hormone made by the thyroid gland is thyroid hormone, also called '[thyroxine](#)', or T4. It is released by the thyroid gland into the bloodstream whenever it is needed by the body. It helps cells work more efficiently and also helps maintain our body temperature. In babies and young children, thyroid hormone is crucial for normal growth and development of the body and brain. Babies who do

not have enough thyroid hormone are often slow to grow, are sluggish, and have learning delays and other specific health problems.

What problems can Hypothyroidism cause?

Hypothyroidism disease is different for each child. Some children with Hypothyroidism have only a few health problems, while other children may have serious complications.

If Hypothyroidism is not treated, a child might develop:

- Coarse, swollen facial features
- Breathing problems
- Hoarse-sounding cry
- Delayed milestones (sitting, crawling, walking, talking)
- Wide, short hands
- Poor weight gain and growth
- [Goiter](#) (enlarged thyroid gland causing a lump in the neck)
- [Anemia](#)
- Slow heart rate
- Fluid build-up under the skin (called [myxedema](#))
- Hearing loss

It is important to follow the doctor's instructions for testing and treatment.

What is the treatment for Hypothyroidism

Hypothyroidism can be treated. The treatment is life-long. Treatment for children with Hypothyroidism can include:

- Thyroid Hormone Replacement
- Regular Monitoring
- Developmental Evaluation

Children with Hypothyroidism should see their regular doctor, a doctor who specializes in Hypothyroidism, and a dietician.

Children with Hypothyroidism can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>

Isovaleric Acidemia (IVA)

(ice-o-va-lair-ic acid-ee-me-ah)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called IVA (eye-vee-ay).

A positive newborn screen does not mean your baby has IVA, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like IVA.

What problems can isovaleric acidemia cause?

IVA is different for each child. Some children with IVA have few health problems, while other children may have very serious complications.

If IVA is not treated, a child might develop:

- Sleepiness
- Feeding problems
- "Sweaty feet" odor
- Seizures
- Stroke
- Mental retardation

It is very important to follow the doctor's instructions for testing and treatment.

What is isovaleric acidemia?

IVA affects an enzyme needed to break down proteins from the food we eat so they can be used for energy and growth. In IVA, the enzyme used to break down proteins is missing or not working properly.

A person with IVA doesn't have enough enzyme to break down protein containing leucine, which causes a substance called isovaleric acid to build up in the body. Isovaleric acid is harmful to the body and causes health problems if it builds up.

IVA is a disorder that is passed on, or inherited, from a child's mother and father. Because IVA is a genetic disease, family members are at risk of having IVA too, even if no one in the family has had it before.

What is the treatment for isovaleric acidemia?

IVA can be treated. The treatment is life-long. Treatment for children with IVA can include:

- Medications to help the body get rid of isovaleric acid and make energy
- Diet low in leucine - a dietician will help you set up the best diet for your child

Children with IVA should see their regular doctor, a doctor who specializes in IVA, and a dietician. Children with IVA can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>

Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)

(long-chain 3-hi-drok-see ay-sill-co-A dee-hi-drodge-in-ace de-fish-in-sea)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called LCHAD (ell-chad). There is another very rare disorder called trifunctional protein deficiency (TFP) that can cause this problem too.

A positive newborn screen does not mean your baby has LCHAD or TFP, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like LCHAD and TFP.

What problems can LCHAD cause?

LCHAD is different for each child. Some children with LCHAD have fewer health problems, while other children may have very serious complications.

If LCHAD is not treated, a child might develop:

Low blood sugar

Sleepiness

Vomiting

Behavior changes (such as crying for no reason)

Seizures

Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is LCHAD?

LCHAD affects an enzyme needed to break down fats in the food we eat so they can be used for energy and growth. In LCHAD, the enzyme used to break down fats is missing or not working properly. A person with LCHAD doesn't have enough enzyme to break down fat into energy. Using stored fat for energy is especially important in

between meals when the baby is not getting new energy from eating food.

LCHAD is a disorder that is passed on, or inherited, from a child's mother and father. Because LCHAD is a genetic disease, family members are at risk of having LCHAD too, even if no one in the family has had it before.

What is the treatment for LCHAD?

LCHAD can be treated. The treatment is life-long. Treatment for children with LCHAD can include:
Frequent meals/snacks and a low fat, high carb diet - a dietician will help you learn what foods can be eaten
Medications to help the body make energy and get rid of harmful toxins
Careful treatment of routine illness

Children with LCHAD should see their regular doctor, a doctor who specializes in LCHAD, and a dietician. Children with LCHAD and TFP can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders
www.rarediseases.org

National Urea Cycle Disorders Foundation
www.nucdf.org

Emory Genetics Lab
<http://genetics.emory.edu/>

Georgia Department of Public Health
<http://health.state.ga.us/programs/nsmscd/>.

Maple Syrup Urine Disease (MSUD) **(ma-pel sir-ap yoor-in di-zez)**

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called MSUD (em-ess-you-dee).

A positive newborn screen does not mean your baby has MSUD, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like MSUD.

What is MSUD?

MSUD affects an enzyme needed to break down proteins from the food we eat, so they can be used for energy and growth. In MSUD, the enzyme used to break down proteins is missing or not working properly. A person with MSUD doesn't have enough enzyme to break down protein containing branched chain amino acids (BCAA). When the body can't break down BCAAs, they build up in the body and cause health problems. The buildup of BCAAs can make the baby's urine smell like pancake syrup.

MSUD is a disorder that is passed on, or inherited, from a child's mother and father. Because MSUD is a genetic disease, family members are at risk of having MSUD too, even if no one in the family has had it before.

What problems can MSUD cause?

MSUD is different for each child. Some children with MSUD have fewer health problems, while other children may have very serious complications.

If MSUD is not treated, a child might develop:

Feeding problems

Weight loss

High-pitched cry

Urine that smells like maple syrup

Sleepiness

Vomiting

Developmental delay

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for MSUD?

MSUD can be treated. The treatment is life-long. Treatment for children with MSUD can include:

Special formula low in BCAAs

Diet low in BCAAs - a dietician will help you set up the best diet for your child

Children with MSUD should see their regular doctor, a doctor who specializes in MSUD, and a dietician. Children with MSUD benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>

Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) **(medium-chain ay-sill co-A dee-hi-drodge-in-ace de-fish-in-sea)**

POSITIVE NEWBORN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called MCAD (em-cad). **A positive newborn screen does not mean your baby has MCAD, but it does mean your baby needs more testing to know for sure.** Your baby's doctor will help arrange for more testing by specialists in disorders like MCAD.

What is MCAD?

MCAD affects an enzyme needed to break down fats in the food we eat, so they can be used for energy and growth. In MCAD, an enzyme used to break down fats is missing or not working properly. A person who has MCAD doesn't have enough enzyme to break down fat into energy. Using stored fat for energy is especially important between meals when the body is not getting new energy from eating food. MCAD is a disorder that is passed on, or inherited, from a child's mother and father. Because MCAD is a genetic disease, family members are at risk of having MCAD too, even if no one in the family has had it before.

What problems can MCAD cause?

MCAD is different for each child. Some children with MCAD have only a few health problems, while other children may have serious complications.

If MCAD is not treated, a child might develop:

- Serious illness (metabolic crisis)
- Sleepiness or little energy
- Behavior changes (such as crying for no reason)
- Irritable mood
- Poor appetite
- Seizures
- Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for MCAD?

There are treatments for children with MCAD, which are life-long. Treatments for children with MCAD can include:

- Frequent meals/snacks and low fat/high carb diet - a dietician will help you set up the best diet for your child
- Medications to help the body make energy and get rid of harmful toxins
- Careful treatment to routine illnesses

Children with MCAD should see their regular doctor, a doctor who specializes in MCAD, and a dietician. Children with MCAD can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

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Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>

Methylmalonic Acidemia (mutase) deficiency (MMA) & Cbl A, B)

(methel-ma-lawn-ic acid-ee-me-ah)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called MMA (em-em-ay). There are several other conditions that can cause a similar positive result on newborn screening.

A positive newborn screen does not mean your baby has MMA, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like MMA.

What is methylmalonic acidemia?

MMA affects enzymes needed to break down proteins and fats from the food we eat so they can be used for energy and growth. There are different kinds of MMA, depending on which enzyme is missing or not working properly. A person with MMA doesn't have enough enzyme to break down protein or fats, so harmful toxins can build up in the body and cause health problems. MMA is a disorder that is passed on, or inherited, from a child's mother and father. Because MMA is a genetic disease, family members are at risk of having MMA too, even if no one in the family has had it before.

What problems can methylmalonic acidemia cause?

MMA is different for each child. Some children with MMA have fewer health problems, while other children have very serious complications.

If MMA is not treated, a child might develop:

- Sleepiness
- Feeding problems
- Muscle weakness
- Seizures
- Stroke
- Poor growth
- Kidney disease
- Mental retardation

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for methylmalonic acidemia?

MMA can be treated. The treatment is life-long. Treatment for children with MMA can include:

Medications to help the body make energy

Vitamin B₁₂

Low-protein diet - a dietician helps families set up the best diet for their child

Children with MMA should see their regular doctor, a doctor who specializes in MMA, and a dietician. Children with MMA can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>.

Multiple Carboxylase Deficiency (MCD) (mul-te-pel car-box-il-ace de-fish-in-sea)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby likely has a disorder called MCD.

A positive newborn screen means your baby likely has MCD and needs more testing to know for sure. Your baby's doctor will help arrange for more testing with specialists in MCD.

What is MCD?

MCD occurs when an [enzyme](#) called "holocarboxylase synthetase" (HCS), is either missing or not working properly. This enzyme's job is to add a vitamin called '[biotin](#)' to other enzymes, so that they can change the food we eat into energy for the body. When the HCS enzyme is not working, certain harmful substances build up in the blood and urine. A number of enzymes are needed to process [protein](#) from the food we eat for use by the body. Problems with one or more of these enzymes can cause an organic acid disorder. People with organic acid disorders cannot break down protein properly. This causes harmful substances to build up in their blood and urine. These substances can affect health, growth and learning.

What problems can MCD cause?

MCD disease is different for each child. Some children with MCD have only a few health problems, while other children may have serious complications. If MCD is not treated, a child might develop:

- Episodes of illness called [metabolic crises](#)
- poor appetite

- vomiting
- extreme sleepiness or lack of energy
- irritability
- low [muscle tone](#) (floppy muscles and joints)
- severe peeling skin rash

It is important to follow the doctor's instructions for testing and treatment.

What is the treatment for MCD

MCD can be treated. The treatment is life-long. Treatment for children with MCD can include:

- Biotin Treatment

Children with MCD should see their regular doctor, a doctor who specializes in MCD, and a dietician. Children with MCD can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>

Phenylketonuria (PKU)

(fee-null-kee-tone-yer-ee-ah)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen shows an increase in a component of protein called phenylalanine (PHE). This result suggests your baby might have a disorder called PKU. There are forms of PKU that can also cause an increase in PHE on newborn screening. These other forms are very rare.

A positive newborn screen does not mean your baby has PKU, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like PKU.

What is PKU?

PKU affects an enzyme needed to break down proteins from the food we eat, so they can be used for energy and growth. In PKU, an enzyme used to break down proteins is missing or not working properly. A person who has PKU doesn't have enough enzyme to break down protein containing phenylalanine (PHE), so harmful toxins can build up in the body. PKU is a disorder that is passed on, or inherited, from a child's mother and father. Because PKU is a genetic disease, family members are at risk of having PKU too, even if no one in the family has had it before.

What problems can PKU cause?

PKU is different for each child. Some children with PKU have only a few health problems, while other children may have serious complications. An infant with PKU might be late in learning to sit, crawl, and stand. Older children may need extra help in school. If PKU is not treated, a child might also develop:

Hyperactivity
Learning disabilities
Seizures
Mental retardation

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for PKU?

There are treatments for children with PKU, which are life-long. Treatments for children with PKU can include:

Special formula low in PHE
Diet low in PHE - a dietician helps families set up the best diet for their child

Children with PKU should see their regular doctor, a doctor who specializes in PKU, and a dietician. Children with PKU can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders
www.rarediseases.org

National Urea Cycle Disorders Foundation
www.nucdf.org

Emory Genetics Lab
<http://genetics.emory.edu/>

Georgia Department of Public Health
<http://health.state.ga.us/programs/nsmscd/>

Propionic Acidemia (PA)
(pro-pe-on-ik acid-ee-me-ah)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby likely has a disorder called PA

A positive newborn screen means your baby likely has PA and needs more testing to know for sure.

Your baby's doctor will help arrange for more testing with specialists in PA

What is PA?

PA occurs when an [enzyme](#) called "propionyl CoA carboxylase" (PCC) is either missing or not working properly. This enzyme's job is to change certain amino acids so the body can use them. When this enzyme is not working, substances called [glycine](#) and [propionic acid](#), along with other harmful substances, build up in the blood and cause problems.

What problems can PA cause?

MCD disease is different for each child. Some children with MCD have only a few health problems, while other children may have serious complications.

If MCD is not treated, a child might develop:

- poor appetite
- vomiting
- irritable mood
- extreme sleepiness or lack of energy
- low [muscle tone](#) (floppy muscles and joints)
- breathing problems
- [seizures](#)
- swelling of the brain
- [stroke](#)
- [coma](#), sometimes leading to death

It is important to follow the doctor's instructions for testing and treatment.

What is the treatment for PA

PA can be treated. The treatment is life-long. Treatment for children with PA can include:

- Low-protein diet
- Medical formula and foods
- Avoid going a long time without food
- Medication
- Regular blood and urine tests
- Liver transplant
- Biotin Treatment

Children with PA should see their regular doctor, a doctor who specializes in PA and a dietician. Children with PA can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

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Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>.

Trifunctional Protein Deficiency

(try-funk-sha-nal pro-ten de-fish-in-sea)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby likely has a disorder called TFP

A positive newborn screen means your baby likely has TFP and needs more testing to know for sure.

Your baby's doctor will help arrange for more testing with specialists in TFP

What is TFP?

TFP deficiency occurs when a group of [enzymes](#), called "trifunctional protein" (TFP), is either missing or not working properly. The job of TFP is to break down certain fats from the food we eat into energy. It also breaks down fat already stored in the body

What problems can TFP cause?

TFP disease is different for each child. Some children with TFP have only a few health problems, while other children may have serious complications.

If TFP is not treated, a child might develop:

- fever
- nausea
- diarrhea
- vomiting
- [hypoglycemia](#)
- increased levels of acidic substances in the blood, called [metabolic acidosis](#)
- breathing problems
- [seizures](#)

- [coma](#), sometimes leading to death

It is important to follow the doctor's instructions for testing and treatment.

What is the treatment for TFP

TFP can be treated. The treatment is life-long. Treatment for children with TFP can include:

- Avoid going a long time without food
- A low fat, high [carbohydrate](#) diet
- MCT oil and L-carnitine
- Avoid heavy exercise and extreme cold.

Children with TFP should see their regular doctor, a doctor who specializes in TFP and a dietician. Children with TFP can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>

Tyrosinemia

(tie-ro-sin-ee-me-ah)

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby likely has a disorder called Tyrosinemia

A positive newborn screen means your baby likely has Tyrosinemia needs more testing to know for sure. Your baby's doctor will help arrange for more testing with specialists in Tyrosinemia

What is Tyrosinemia?

Tyrosinemia occurs when an enzyme, called fumarylacetoacetase (FAH), is either missing or not working properly. When FAH is not working, it cannot break down tyrosine. Tyrosine and other harmful substances then build up in the blood. One of these substances is called [succinylacetone](#). When it builds up in the blood, it causes serious liver and kidney damage. It may also cause episodes of weakness or pain.

What problems can Tyrosinemia cause?

Tyrosinemia disease is different for each child. Some children with Tyrosinemia have only a few health problems, while other children may have serious complications.

If Tyrosinemia is not treated, a child might develop:

- diarrhea and bloody stools
- vomiting
- poor weight gain
- extreme sleepiness
- irritability
- “cabbage-like” odor to the skin or urine
- enlarged liver
- yellowing of the skin
- tendency to bleed and bruise easily
- swelling of the legs and abdomen
- [rickets](#), a bone thinning condition
- delays in walking
- pain or weakness, especially in the legs
- breathing problems
- rapid heartbeat
- [seizures](#)
- [coma](#), sometimes leading to death

It is important to follow the doctor’s instructions for testing and treatment.

What is the treatment for Tyrosinemia

Tyrosinemia can be treated. The treatment is life-long. Treatment for children with Tyrosinemia can include:

- Medication
- Medical Formula
- Low- tyrosine/phenylalanine diet
- Blood, urine, and other test
- Liver transplantation

Children with Tyrosinemia should see their regular doctor, a doctor who specializes in Tyrosinemia and a dietician.

Children with Tyrosinemia can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

Very Long-Chain Acyl-CoA Dehydrogenase Deficiency **(VLCAD)** **(long-chain ay-sill co-A dee-hi-drodge-in-ace de-fish-in-sea)**

POSITIVE NEWBORN SCREEN

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby likely has a disorder called VLCAD.

A positive newborn screen means your baby likely has VLCAD needs more testing to know for sure. Your baby's doctor will help arrange for more testing with specialists in **VLCAD**

What is VLCAD?

VLCAD occurs when an [enzyme](#), called "very long chain acyl-CoA dehydrogenase" (VLCAD) is either missing or not working properly. This enzyme's job is to break down certain fats from the food we eat and turn it into energy. It also breaks down fat already stored in the body. When the VLCAD enzyme is missing or not working, the body cannot break down fat for energy and must rely solely on glucose. Although glucose is a good source of energy, there is a limited amount available. Once the glucose has been used up, the body tries to use fat without success. This leads to low blood sugar, called [hypoglycemia](#), and to the buildup of harmful substances in the blood.

What problems can VLCAD cause?

VLCAD disease is different for each child. Some children with VLCAD have only a few health problems, while other children may have serious complications.

If VLCAD is not treated, a child might develop:

- extreme sleepiness
- behavior changes
- irritable mood
- poor appetite
- fever
- nausea
- diarrhea
- vomiting
- hypoglycemia
- breathing problems
- [seizures](#)

- [coma](#), sometimes leading to death

It is important to follow the doctor's instructions for testing and treatment.

What is the treatment for VLCAD

VLCAD can be treated. The treatment is life-long. Treatment for children with VLCAD can include:

- Avoid going a long time without food
- Diet
- MCT oil and L-carnitine
- Avoid prolonged exercise or exertion

Children with VLCAD should see their regular doctor, a doctor who specializes in VLCAD and a dietician. Children with VLCAD can benefit from prompt and careful treatment.

Resources for parents

National Organization for Rare Disorders

www.rarediseases.org

National Urea Cycle Disorders Foundation

www.nucdf.org

Emory Genetics Lab

<http://genetics.emory.edu/>

Georgia Department of Public Health

<http://health.state.ga.us/programs/nsmscd/>