Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

What is MCAD?
MCAD is a type of fatty acid oxidation disorder. People with MCAD have problems breaking down fat into energy for the body.

What Causes MCAD?
Enzymes help start chemical reactions in the body. MCAD happens when an enzyme called “medium chain acyl-CoA dehydrogenase” is either missing or not working. This enzyme breaks down certain fats in the food we eat into energy. It also breaks down fat already stored in the body.

What Symptoms or Problems Occur with MCAD?
[Symptoms are something out of the ordinary that a parent notices.]
MCAD can cause bouts of illness called Metabolic Crises. Children with MCAD often show symptoms for the first time between 3 months and 2 years of age. Some of the first signs of a Metabolic Crisis are:

- too much sleepiness
- behavior changes
  (such as crying for no reason)
- irritable mood
- poor appetite

If a Metabolic Crisis is not treated, a child with MCAD can develop:

- breathing problems
- seizures
- mental retardation
- cerebral palsy
- coma, sometimes leading to death

What is the Treatment for MCAD?
The following treatments are often used for children with MCAD:

1. Do not go a long time without food – Babies and young children with MCAD need to eat often to avoid low blood sugar or a Metabolic Crisis. They should not go without food for more than 4 to 6 hours. Some babies need to eat even more often. It is important that babies be fed during the night. They need to be woken to eat if they do not wake up on their own. Young children with MCAD may need to have a starchy snack (such as bread, cereal, rice) before bed and another during the night. They may need another snack first thing in the morning. Your dietitian can give you ideas for good night-time snacks. Dietitians know what are the correct foods to eat. Most teens and adults with MCAD can go without food for up to 12 hours without problems when they are well. They need to continue the other treatments for life.

2. Diet – Sometimes a low-fat, high carbohydrate diet (such as vegetables, fruits, grains) is advised. Your dietitian can create a food plan with the right type and amount of fat your child needs. Ask your doctor whether or not your child needs to have any changes in his or her diet.

3. L-carnitine – L-carnitine (Carnitor) may be prescribed for some children. This is safe and natural and helps body cells make energy. It also helps the body get rid of harmful wastes.

Things to Remember
Always call your doctor when your child has any of the following:

- poor appetite
- low energy or too much sleepiness
- vomiting
- diarrhea
- an infection
- a fever

People with MCAD need to eat extra starchy foods and drink more fluids during any illness – even if they don’t feel hungry – or they could develop low blood sugar or a Metabolic Crisis. Children who are sick often don’t want to eat. If they won’t or can’t eat, they may need to be treated in the hospital to prevent problems.
3-Methylcrotonyl CoA Carboxylase Deficiency (3MCC)

What is 3MCC?

3MCC is a type of organic acid disorder. People with this condition can’t break down an amino acid called leucine from the food they eat.

What Causes 3MCC?

Enzymes help start chemical reactions in the body. 3MCC happens when an enzyme called “3-methylcrotonyl CoA carboxylase (3MCC)” is missing or doesn’t work right. This enzyme helps break down leucine. All foods with protein contain leucine. Harmful matter can build up in the blood and cause problems when someone with 3MCC eats protein.

What Symptoms or Problems Occur with 3MCC?

[Symptoms are something out of the ordinary that a parent notices.]

Symptoms of 3MCC vary. Many people with 3MCC have no symptoms. Others have periods of illness called Metabolic Crises. Some of the first signs are:

- poor appetite
- too much sleepiness or lack of energy
- muscle weakness
- irritable mood
- other behavior changes (such as crying for no reason)
- vomiting

If a Metabolic Crisis is not treated, a child with 3MCC might develop:

- breathing problems
- seizures
- liver failure
- coma, sometimes leading to death

Later problems can include:

- low muscle tone
- weakness
- lack of energy
- poor growth

What is the Treatment for 3MCC?

These treatments are sometimes used for babies and children with 3MCC:

1. Low-leucine diet - Most foods in a low-leucine diet will be carbohydrates (such as bread, cereal, noodles, fruits, vegetables). High-protein foods to limit or avoid include:
   - milk and milk products
   - meat and poultry
   - fish
   - eggs
   - dried beans and peas
   - nuts/peanut butter

2. Medical foods and formula – There are special medical foods available for people with 3MCC. These foods include special low-protein flours, noodles, and rice. Some children also need a special leucine-free formula. A dietitian will tell you how to use these foods and formula. Dietitians know what are the right foods to eat.

3. Medications – The doctor may prescribe L-carnitine for your child. This is safe and natural and helps cells make energy. It also helps the body get rid of harmful wastes.

Things to Remember

Minor illnesses such as a cold or flu can cause a Metabolic Crisis in some children with 3MCC. You may need to call your doctor right away when your child has any of the following:

- loss of appetite
- vomiting
- infection or other illness
- fever
- diarrhea

Some children with 3MCC need to eat more starchy foods (such as bread, cereal, rice) and drink more fluids when they are sick - even if they are not hungry – to avoid a Metabolic Crisis.
Argininosuccinic Acidemia (ASA)

What is ASA?
ASA is a type of amino acid disorder. People with this condition can’t remove ammonia from the body. Ammonia is a harmful substance. It is made when the body breaks down protein and amino acids for use by the body.

What Causes ASA?
ASA is a “urea cycle disorder” (UCD). ASA happens when an enzyme called “argininosuccinic acid lyase” (ASAL) is missing or not working. Enzymes help start chemical reactions in the body. Ammonia builds up in the blood when there is a problem with the ASAL enzyme. Too much ammonia in the blood can cause brain damage. It can also cause death if not treated.

What Symptoms or Problems Occur with ASA?
[Symptoms are something out of the ordinary that a parent notices.]

There are two kinds of ASA. The severe form starts in babies. They are healthy when born, but soon show symptoms of high ammonia levels. The milder form of ASA starts in childhood.

Some of the first symptoms of high ammonia are:
- poor appetite
- too much sleepiness or no energy
- irritable mood
- vomiting

If not treated, high ammonia can cause:
- muscle weakness
- breathing problems
- problems staying warm
- seizures
- swelling of the brain
- coma, sometimes leading to death

The milder form can also cause mental retardation, seizures, a large liver, and skin and hair problems.

What Is the Treatment for ASA?
The following treatments are often used for babies and children with ASA:

1. **Low-protein diet and/or special medical foods and formula** – The best treatment is a very low-protein diet. There are medical foods such as special low-protein flours, noodles, and rice available. A dietitian will make a food plan for your child. Dietitians know what are the right foods to eat. Your child will need to eat a low-protein diet for life. The doctor or dietitian may give your baby a special formula that has the right nutrients and amino acids.

2. **Medication** – The doctor might prescribe arginine supplements for your child. Other medicines may be used to prevent high ammonia.

3. **Blood tests** – Regular blood tests will check your child’s amino acid and ammonia levels.

Things to Remember
Children with high ammonia often need to be treated in the hospital. Call your doctor right away if your child has any of the following:

- loss of appetite
- low energy or too much sleepiness
- vomiting
- fever
- bad headache
- infection or illness
- behavior or personality changes (such as crying for no reason)
- problems walking or balancing
What is BIOT?
BIOT is an inherited disorder. BIOT happens when the body can’t use a vitamin called biotin. People with BIOT do not have enough biotinidase activity. This condition is treatable.

What Causes BIOT?
Low biotinidase activity keeps carboxylases from using biotin. The body collects harmful matter when carboxylases can’t break down nutrients in the right way. Children with BIOT need extra biotin, or health problems usually result.

What Symptoms or Problems Occur with BIOT?
Early symptoms of untreated BIOT are:
- seizures
- poor muscle tone
- vision problems
- poor coordination
- delay in development
- hearing loss
- skin abnormalities such as:
  - hair loss
  - rash
  - infection

Problems that can occur with age are:
- motor limb weakness
- loss of body control and/or feeling
- poor vision

What is the Treatment for BIOT?
Medication – Newborns with BIOT rarely have symptoms if they are treated right away. Children with symptoms improve when they take biotin every day. People with severe BIOT should take biotin all of their lives.

Diet – Avoid raw eggs. Well-cooked eggs are safe to eat.

Things to Remember
Some infants with BIOT don’t have symptoms. They often don’t have problems if they get early treatment. They just need to take biotin every day. They also need to get regular medical checkups.

Children usually get better right away when treated soon after they show symptoms. Some children might continue to have problems. These include hearing loss, trouble seeing, or developmental problems.

Your doctor can explain more about your child’s biotin treatment.
Beta Ketothiolase Deficiency (BKD)

What is BKD?
BKD is a type of organic acid disorder. People with BKD can’t break down an amino acid called isoleucine from the food they eat.

What Causes BKD?
Enzymes help start chemical reactions in the body. BKD happens when an enzyme called “mitochondrial acetoacetyl-CoA thiolase” (MAT) is missing or not working. This enzyme helps break down the amino acid isoleucine. All foods with protein contain isoleucine. Harmful matter can build up in the blood and cause problems when someone with BKD eats protein.

What Symptoms or Problems Occur with BKD?

[Symptoms are something out of the ordinary that a parent notices.]

BKD can cause periods of illness called Metabolic Crises. Some of the first symptoms of a Metabolic Crisis are:

- too much sleepiness or lack of energy
- vomiting and diarrhea
- fever
- poor appetite
- ketones in the urine

Later problems can include:

- mental retardation
- dystonia (muscle spasms)
- enlarged heart
- low blood platelets
- higher risk for infections
- not growing well
- kidney problems

What is the Treatment for BKD?
The treatments often recommended for babies and children with BKD are:

1. Medication – The doctor may prescribe L-carnitine for your child. This is safe and natural and helps the body make energy. It also helps the body get rid of harmful wastes.

2. Do not go a long time without food – Some babies and young children can have a Metabolic Crisis if they don’t eat often enough. Your doctor will tell you how to space your child’s meals. Most healthy children over the age of 10 can go without food for up to 12 hours without problems.

3. Low-protein diet – Some children will need to eat a low-protein diet. High-protein foods to be limited include:

- milk and milk products
- meat and poultry
- fish
- eggs
- dried beans
- nuts/peanut butter

Children with BKD need some protein to grow properly. A dietitian can make a food plan for your child. Dietitians know what are the correct foods to eat.

Things to Remember
Even minor illness such as a cold or flu can lead to a Metabolic Crisis. Call your doctor right away when your child has any of the following:

- loss of appetite
- vomiting
- diarrhea
- infection or illness
- fever

An ill child with BKD needs more liquids, starchy foods (such as rice, cereal, bread), and sugars to avoid a Metabolic Crisis. When your child is ill, you will also need to limit protein.

Children with symptoms of a Metabolic Crisis need to be treated in a hospital.
### What is CAH?
Babies with Congenital Adrenal Hyperplasia (CAH) are born with enlarged adrenal glands. The adrenal glands of these babies can't make enough of a hormone called cortisol — the main hormone in adrenal glands. The adrenal glands get too big when they try to make the right amount of this hormone.

### What Causes CAH?
The adrenal glands make chemical messengers called hormones. The hormones they make are cortisol (hydrocortisone), aldosterone (salt-retaining hormone), and androgens (male sex hormones). CAH happens when certain enzymes in the adrenal glands are missing. Enzymes start chemical reactions in the body. The missing enzymes cause the glands to make too little of the cortisol and aldosterone hormones and too much of the male-like hormones. CAH is an inherited disorder. Both parents carry the gene for CAH.

### What Symptoms or Problems Occur with CAH?

*Symptoms are something out of the ordinary that a parent notices.*

There are three main forms of CAH:
- severe salt-wasting
- non-salt wasting
- milder form

An infant with the severe salt-wasting form may have one or more of these symptoms in the first weeks of life:
- vomiting
- poor weight gain
- poor feeding
- sleepiness
- diarrhea
- dehydration (loss of fluids)

Newborns with the non-salt wasting form of CAH usually don’t get severely sick. A girl’s genitals may look more like a boy’s. Other symptoms of the non-salt wasting form develop with age. Both boys and girls may have:
- rapid growth in early childhood
- early sexual development
- early pubic hair growth

Children with the milder form of CAH show symptoms anytime between early childhood and puberty. They might grow more quickly or grow pubic hair early. Girls at puberty usually have:
- excess body hair growth
- acne
- irregular periods
- sometimes infertility

### What is the Treatment for CAH?

**Medications** – CAH is treated with two types of hormone medicines. Both boys and girls with CAH need to take a cortisol-like hormone pill. This will prevent the body from making too much of the male-like hormone. Children with CAH take the pills two or three times a day. Children with salt-wasting CAH may also take a salt-saving hormone pill, plus salt tablets. Your doctor may tell you to add salt to your baby's formula. A cortisol shot may be necessary if your child gets sick.

### Things to Remember
Children with CAH must take hormone pills all of their lives. Your doctor will check your child’s height, weight, and blood pressure. He or she may x-ray the wrist to look at the bone age and may do a blood test to check hormone levels. Your doctor will prescribe the hormone medicine after testing. He or she may change the amount as your child grows or gets sick or hurt.
Congenital Hypothyroidism
(CH)

What Is CH?
CH is a condition in which the thyroid gland doesn’t make enough thyroid hormone. The thyroid gland is in the neck.

What Causes CH?
CH happens when the thyroid gland doesn’t make any or enough hormones. This mainly occurs because the thyroid gland didn’t develop, or because it developed in a different place in the body. No one knows why the thyroid gland doesn’t develop all the way, or why it doesn’t develop where it should.

What Symptoms or Problems Occur with CH?
(Symptoms are something out of the ordinary that a parent notices.)

There are different symptoms of CH in babies. The most obvious are:

- Skin: Some CH babies have newborn jaundice (a yellow color) longer than usual. Their skin may look pale or blotchy.
- Appetite and Digestion: CH babies may not want to feed. It might be hard to keep them awake during feedings. They may be very constipated.
- Growth: CH newborns are often large. They usually have poor growth and weight gain if they are not treated.
- Circulation: Babies with CH may have a slow heart rate and low blood pressure. Their hands and feet may be cool to the touch. This is because of poor circulation.
- Activity and Development: Babies with CH are often quiet and don’t cry much. They may not be interested in sights or sounds around them. They may sleep for a long time, then have to be woken up for feeding. They may feel “floppy” when they are picked up.

What is the Treatment for CH?
Medications – The doctor will prescribe synthetic (man-made) thyroid hormone to replace your child’s missing thyroid hormone. The medicine acts just like the natural hormone made in the body. The synthetic thyroid hormone doesn’t cause other problems when the right amount is given.

Things to Remember
Babies will have normal growth and development when they get their thyroid medicine regularly. Your baby has the same chance for a normal life as any other baby.
Newborn Screening FACT Sheet

Citrullinemia (CIT)

What is CIT?
CIT is a type of amino acid disorder. People with CIT can’t rid the body of ammonia. It is made when the body breaks down protein and amino acids.

What Causes CIT?
Enzymes help start chemical reactions in the body. CIT is a condition called “urea cycle disorder.” It happens when an enzyme called “argininosuccinic acid synthetase” (ASAS) is either missing or doesn’t work right. ASAS helps break down amino acids. It also removes ammonia from the body. The amino acid citrulline builds up in the blood when ASAS doesn’t work. Ammonia also builds up. Too much ammonia can cause brain damage. It can cause death if untreated.

What Symptoms or Problems Occur with CIT?
[Symptoms are something out of the ordinary that a parent notices.]

Some of the first symptoms of high ammonia are:
- poor appetite
- too much sleepiness or lack of energy
- irritable mood
- vomiting

If untreated, high ammonia can cause:
- muscle weakness
- breathing problems
- problems staying warm
- seizures
- swelling of the brain
- coma, sometimes leading to death

What is the Treatment for CIT?

1. Low-protein diet and/or special medical foods and formula – The best treatment for CIT is a very low-protein diet (avoid meat, fish, eggs, milk products, nuts and beans). There are medical foods such as special low-protein flours, noodles, and rice available. A dietitian will make a food plan for your child. Dietitians know what the right foods to eat. The doctor or dietitian may give your baby a special formula with the right nutrients and amino acids. People with CIT should follow their food plan for life.

2. Medication – Medications can also rid the body of ammonia. Children with CIT take these by mouth or feeding tube.

3. Blood tests – Regular blood tests will check your child’s amino acid and ammonia levels.

Things to Remember
Call your doctor right away if your child has any of the following:
- loss of appetite
- low energy or too much sleepiness
- vomiting
- fever
- infection or illness
- behavior or personality changes (such as crying for no reason)
- problems walking or balancing
- bad headache

Children with high ammonia often need to be treated in the hospital.
Carnitine Uptake Deficiency (CUD) or Carnitine Transport Deficiency (CTD)

What is CUD?
CUD is a kind of fatty acid oxidation disorder. People with CUD have problems breaking down fat into energy for the body.

What Causes CUD?
Enzymes help start chemical reactions in the body. CUD happens when an enzyme called “carnitine transporter” is either missing or not working right. This enzyme helps carry something called carnitine into our cells. Carnitine helps the body make energy from fats in food. It also helps us to use the fat that is already stored in the body.

What Symptoms or Problems Occur with CUD?
[Symptoms are something out of the ordinary that a parent notices.]

Babies with CUD first show signs of it between birth and age three. CUD can cause periods of illness called Metabolic Crises. Some of the first symptoms of a Metabolic Crisis are:
- too much sleepiness
- behavior changes (such as crying for no reason)
- irritable mood
- poor appetite

If a Metabolic Crisis is not treated, a child with CUD can develop:
- breathing problems
- swelling of the brain
- seizures
- coma, sometimes leading to death

Repeated Metabolic Crises can cause brain damage. This can cause learning problems or mental retardation. CUD can also cause an enlarged heart and weak muscles.

What is the Treatment for CUD?
The following treatments are often used for children with CUD:

1. **L-carnitine** – The doctor usually prescribes the life-long use of L-carnitine. This is safe and natural and helps the body make energy. It also helps the body get rid of harmful wastes. L-carnitine can get rid of heart problems and muscle weakness in children with CUD.

2. **Do not go a long time without food** – Babies and young children with CUD should eat more often to avoid problems. They should not go without food for more than 4 to 6 hours. Some babies may need to eat even more often than this. It is important that babies be fed during the night. They need to be woken to eat if they do not wake up on their own.

3. **Diet** – Sometimes a low-fat, high carbohydrate diet (such as fruits, vegetables, bread, noodles) is used in addition to L-carnitine. Any changes in the diet should be made by a dietitian. Dietitians know what are the right foods to eat. Ask your doctor whether your child needs to have any changes in his or her diet.

Things to Remember
Always call your doctor when your baby has any of the following:
- poor appetite
- low energy or too much sleepiness
- vomiting
- diarrhea
- an infection
- a fever
- continued muscle pain or weakness

Babies with CUD need to eat extra starchy food (such as rice, cereal, bread) and drink more fluids when they’re sick - even if they don’t feel hungry – or they could have a Metabolic Crisis. If they won’t or can’t eat, they may need to be treated in the hospital to prevent serious health problems.
Glutaric Acidemia, Type 1 (GA1)

What is GA1?
GA1 is a type of organic acid disorder. People with GA1 can't break down the amino acids lysine and tryptophan from the foods they eat.

What Causes GA1?
Enzymes help start chemical reactions in the body. GA1 happens when an enzyme called "glutaryl-CoA dehydrogenase" is missing or not working. This enzyme breaks down glutaric acid. Glutaric acid is made when the body breaks down lysine, hydroxylysine and tryptophan. When a child with GA1 eats food with these amino acids, glutaric acid builds up in the blood. All foods with protein contain these amino acids.

What Symptoms or Problems Occur with GA1?
[Symptoms are something out of the ordinary that a parent notices.]
Newborns with GA1 are usually healthy, but many are born with a large head. Other symptoms usually start between 2 months and 4 years of age.

GA1 causes periods of severe illness called Metabolic Crises. Some early signs of a Metabolic Crisis are:
- poor appetite
- too much sleepiness or lack of energy
- irritable mood
- feeling jittery
- nausea
- vomiting
- low muscle tone (floppy muscles and joints)
- weak muscles

If untreated, more symptoms can follow:
- tics or muscle spasms
- rigid muscles
- jerking movements of the arms and legs
- poor coordination and balance
- high levels of acids in the blood
- seizures
- brain swelling, or blood in the brain
- coma, sometimes leading to death

What is the Treatment for GA1?
Treatments for babies and children with GA1 are:

1. Medication – The doctor may prescribe riboflavin for your child. This is a vitamin that helps the body use protein. It helps remove glutaric acid from the blood. The doctor might also prescribe L-carnitine. This is safe and natural and helps the body make energy. Don’t use any medicine without checking with your doctor.

2. Food plan, including medical foods and formula – Most children need to eat foods low in lysine and tryptophan. The diet often includes special medical foods and formulas. The following foods should not be eaten at all or limited:
   - milk, cheese and other milk products
   - meat and poultry
   - fish
   - eggs
   - dried beans and peas
   - nuts and peanut butter

3. Blood tests – Regular blood tests will measure your child’s amino acid levels.

Things to Remember
Minor illness such as a cold or the flu can cause a Metabolic Crisis in babies and children with GA1. Call your doctor right away when your child has any of the following:
- loss of appetite
- low energy or too much sleepiness
- vomiting
- fever
- infection or illness
- behavior or personality changes (such as crying for no reason)

Sick children often don’t feel hungry. If they can’t eat or show signs of a Metabolic Crisis, they may need to be treated in the hospital.
What is GALT?
GALT is a rare, inherited problem. It is caused when the body can’t break down galactose. Galactose is a sugar found in milk and milk products.

What Causes GALT?
Breast milk and most infant formulas have a sugar called lactose. The body breaks lactose down into sugars called glucose and galactose. Galactose must be broken down more before the body can use it for energy. An enzyme called galactose-1-phosphate uridyl transferase helps do this. Enzymes help start chemical reactions in the body. Most people with GALT don’t have this special enzyme. This causes galactose to build up in the body.

What Symptoms or Problems Occur with GALT?
[Symptoms are something out of the ordinary that a parent notices.]
High levels of galactose poison the body and cause these serious problems:
- swollen liver
- kidney failure
- stunted growth and mental retardation
- cataracts in the eyes

Children and young adults treated for GALT may still have problems over the years with:
- speech
- language
- hearing
- clumsiness with hands
- bleeding in the gel-like part of the eye
- tremors (shaking)
- stunted growth
- learning disabilities

What is the Treatment for GALT?

Special Diet – The treatment for GALT is to limit galactose and lactose from the diet for life. All milk and all foods that have milk in them must not be used at all. This includes any kind of milk, such as cow’s milk, goat’s milk, and human breast milk. Your child should also not eat dairy products like butter, cheese, and yogurt. Other foods with small amounts of milk products must also not be eaten. These include foods with whey, casein, and curds.

Things to Remember
Children with GALT should be in the care of a doctor who specializes in the treatment of GALT. You will also have a dietitian who will teach you about special diets for your child. Dietitians know what are the right foods to eat.

Read labels carefully when you shop for your child’s food. Many prepared foods have hidden ingredients that contain galactose.

Many medicines contain fillers that include galactose. It is important to ask the doctor and pharmacist about this for any medicines prescribed for your child.
What is HCY?
HCY is a type of amino acid disorder. People with HCY can’t break down an amino acid called methionine from the food they eat.

What Causes HCY?
Enzymes help start chemical reactions in the body. HCY happens when an enzyme called “cystathionine beta-synthase” (CBS) is missing or not working right. This enzyme breaks down methionine. When the CBS enzyme is not working right, methionine and homocystine (another amino acid) build up in the blood and cause problems.

What Symptoms or Problems Occur with HCY?
Symptoms are something out of the ordinary that a parent notices.

Babies look healthy and normal at birth. If untreated, HCY can cause growth and learning delays. It can also affect the eyes, bones, heart, and blood vessels.

Growth, learning and behavior – Delays in growth and learning are often noticed between ages one and three. Common problems include:
- poor growth
- problems gaining weight
- delays in crawling, walking, and talking
- behavior and emotional problems (such as crying for no reason)
- serious learning disabilities or mental retardation

Eyes – Children with HCY usually start to develop severe nearsightedness after age one. If this isn’t treated, the lens of the eye can get loose and move out of place. This is called “lens dislocation.” It often happens between ages two and eight. Glaucoma is a condition caused by increased eye pressure. This can happen over time if the lens dislocation is not treated. Untreated glaucoma can cause blindness.

Bones and skeleton – Teens and adults with HCY are often tall and slender. They may have very long arms, legs, and fingers. About half have thinning of the bones, called osteoporosis. Some children will have muscle weakness, especially in the legs.

Heart and blood vessels – HCY can cause blood clots resulting in heart disease or stroke if not treated. Stroke and heart disease are the main causes of early death in people with untreated HCY.

Other – Children who are not treated often have pale hair and skin. Some will have periods of pancreatitis (inflammation of the pancreas gland), which causes severe pain.

What is the Treatment for HCY?
Your baby’s primary doctor will work with a metabolic doctor and dietitian to care for your child. Dietitians know what are the right foods to eat.

The following are treatments often used for children with HCY:
- Low-methionine diet
- Special medical formula
- Blood and urine tests
  - Vitamin B6
  - Betaine
  - Vitamin B12
  - Folic Acid
  - L-cystine

Things to Remember
With treatment all life long, many children have normal growth and intelligence. Treatment may lower the chance for blood clots, heart disease, and stroke. Treatment also lessens the chance of eye problems. Even when treated, some people still develop lens dislocation. This can often be fixed by surgery or other methods.

Children who begin treatment later in life may have mental retardation and behavior problems.
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)

What is HMG?
HMG is a type of organic acid disorder. People with this condition have problems breaking down an amino acid called leucine from the food they eat.

What Causes HMG?
Enzymes help start chemical reactions in the body. HMG happens when an enzyme called “HMG CoA lyase” is either missing or not working well. This enzyme has two jobs. The first is to help break down leucine. All foods with protein contain leucine. The second job is to help the body make something called “ketone bodies” from stored fat.

What Symptoms or Problems Occur with HMG?
[Symptoms are something out of the ordinary that a parent notices.]
HMG causes periods of illness called Metabolic Crises. Some of the first symptoms are:
- poor appetite
- too much sleepiness or lack of energy
- irritable mood/behavior changes (such as crying for no reason)
- muscle weakness

Other problems then follow:
- fever
- diarrhea
- vomiting
- hypoglycemia (low blood sugar)
- increased levels of acid matter in the blood, called metabolic acidosis
- high levels of ammonia in the blood
- enlarged liver

If a Metabolic Crisis is not treated, a child with HMG can develop:
- breathing problems
- seizures
- coma, sometimes leading to death

When not treated, many babies die during their first Metabolic Crisis. Babies who live may have repeated Metabolic Crises that can cause brain damage. This can result in life-long learning problems or mental retardation and a seizure disorder.

Other problems can include:
- Heart enlargement
- Pancreatitis (inflamed pancreas gland)
- Hearing loss
- Vision problems

What is the Treatment for HMG?
The following treatments are often used for babies and children with HMG:

1. Do not go a long time without food – Babies and young children need to eat often to avoid a Metabolic Crisis. Most babies should not go without food for more than 4 to 6 hours. Some babies may need to eat more often. It is important to feed babies during the night. They may need to be woken to eat if they don’t wake up on their own.

2. Low-leucine diet, including medical foods and formula – A food plan low in leucine that limits fat and protein is often part of the treatment. High-protein and high-fat foods that your child may need to limit or not eat at all include:
- milk and milk products
- meat and poultry
- fish
- eggs
- dried beans and peas
- nuts and peanut butter
- butter, margarine, oil, lard, and foods made with these fats

3. Medications – Taking L-carnitine may help some children. This is safe and natural and helps the body make energy. Use only the form prescribed by your doctor.

Things to Remember
Even minor illnesses such as a cold or the flu can cause a Metabolic Crisis. Call your doctor right away when your child has any of the following:
- loss of appetite
- vomiting and diarrhea
- infection or illness
- fever

Children who are sick often don’t want to eat. If they can’t eat, or if they show signs of a Metabolic Crisis, they may need to be treated in the hospital.
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)

What is LCHAD?
LCHAD is a type of fatty acid oxidation disorder. People with LCHAD have problems breaking down fat into energy for the body.

What Causes LCHAD?
Enzymes help start chemical reactions in the body. LCHAD happens when an enzyme called “long chain 3-hydroxyacyl-CoA dehydrogenase” is either missing or not working. This enzyme breaks down certain fats from the food we eat into energy. It also breaks down fat already stored in the body.

What Symptoms or Problems Occur with LCHAD?
[Symptoms are something out of the ordinary that a parent notices.]
LCHAD can cause mild problems in some people and more serious problems in others. Babies and children with LCHAD usually begin to show symptoms sometime from birth through age two. LCHAD causes periods of low blood sugar.

The first symptoms of low blood sugar are:
- extreme sleepiness or tiredness
- weakness
- nausea
- vomiting
- feeling irritable or jittery
- behavior changes (such as crying for no reason)

If low blood sugar is not treated, a child with LCHAD can develop:
- breathing problems
- swelling of the brain
- seizures
- coma, sometimes leading to death

Symptoms often show up after having nothing to eat for more than a few hours. They also show up when a child with LCHAD gets sick or has an infection. Nerve problems and vision problems can happen later.

What is the Treatment for LCHAD?
The following treatments are often used for children with LCHAD:

1. Do not go a long time without food – Babies and young children with LCHAD need to eat often to avoid low blood sugar. They should not go without food for more than 4 to 6 hours. Some babies need to eat even more often. It is important that babies be fed during the night.

   Young children with LCHAD should have a starchy snack (such as bread, cereal, rice) before bed and another during the night. They need another snack first thing in the morning. Raw cornstarch mixed with water, milk, or other drink is a good source of long-lasting energy. Your dietitian can give you ideas for good night-time snacks. Dietitians know what are the correct foods to eat.

2. Diet – Sometimes a low-fat, high-carbohydrate (such as vegetables, bread, fruits) diet is advised. People with LCHAD cannot use certain building blocks of fat called “long chain fatty acids.” A dietitian can help create a food plan low in these fats.

3. MCT oil, L-carnitine and other supplements – People with LCHAD often use MCT oil. This special oil has medium chain fatty acids. It can be used in small amounts for energy. Doctors prescribe L-carnitine for some children. This is safe and natural and helps body cells make energy. It also helps the body get rid of harmful wastes. Some doctors suggest taking DHA. This may help prevent loss of eyesight.

Things to Remember
Always call your doctor when your child has any of the following:
- poor appetite
- low energy or too much sleepiness
- vomiting
- diarrhea
- an infection
- a fever
- continuing muscle pain or weakness
- reddish-brown color to the urine

Children with LCHAD need to eat extra starchy food and drink more fluids during any illness.
 Isovaleric Acidemia  
(IVA)

What is IVA?  
Enzymes help start chemical reactions in the body. IVA happens when an enzyme called “isovaleryl-CoA dehydrogenase” is missing or not working well. This enzyme helps break down harmful “isovaleric acid.” This acid builds up in the blood and causes problems when a child with IVA eats food with leucine. Leucine is in all foods that have protein (such as meat, beans, peanut butter, milk).

What Causes IVA?  
People with IVA have a pair of genes that don’t work as they should. These genes cause the “isovaleryl-CoA dehydrogenase” enzyme to not work well or not be made at all.

What Symptoms or Problems Occur with IVA?  
[Symptoms are something out of the ordinary that a parent notices.]

Babies with IVA seem healthy at birth. Symptoms often start between one day and two weeks of age. IVA causes periods of illness called Metabolic Crises. Some of the first signs are:

- poor appetite
- too much sleepiness, low energy
- vomiting
- feeling cold
- “sweaty feet” odor

If a Metabolic Crisis is not treated, a child with IVA may develop:

- breathing problems
- seizures
- strokes
- mental retardation
- coma, sometimes leading to death

The less severe kind of IVA shows up later in childhood. Some problems may include:

- poor growth
- learning difficulties

What is the Treatment for IVA?  
Early treatment prevents Metabolic Crises and related problems. Treatment should start as soon as you know your child has IVA. Treatment usually lasts all life long. Treatment often includes:

1. Low-leucine diet, medical foods and formula – Most children need to eat foods low in leucine (such as vegetables and fruit). Special medical foods and formulas are usually part of the diet. You will get a food plan that has the right amount of protein and nutrients to keep your child healthy. Your child should continue a special food plan for life. High-protein foods your child should limit or not eat include:
   - milk and milk products
   - meat and poultry
   - fish
   - eggs
   - dried beans and peas
   - nuts and peanut butter

2. Medications – The doctor may prescribe the amino acid Glycine to help the body get rid of isovaleric acid. This can help prevent Metabolic Crises in children with IVA. L-carnitine may also help some children. This is safe and natural and helps the body make energy. Only use the kind your doctor tells you to use. Do not use any medication or supplement without checking with your doctor.

Things to Remember  
Even minor illnesses such as a cold or the flu can cause a Metabolic Crisis. Call your doctor right away when your child has any of the following:

- loss of appetite
- vomiting
- diarrhea
- infection or illness
- fever

Children with IVA need to eat more starchy foods (bread, cereal, rice, noodles) and drink more fluids when they are ill - even if they’re not hungry – or they could have a Metabolic Crisis. They also need to not eat protein foods when they are sick. If they can’t eat, or if they show signs of a Metabolic Crisis, they may need to be treated in the hospital.
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

What is VLCAD?
VLCAD is a type of fatty acid oxidation disorder. People with VLCAD can’t break down certain types of fat into energy for the body.

What Causes VLCAD?
Enzymes help start chemical reactions in the body. VLCAD happens when an enzyme called “very long chain acyl-CoA dehydrogenase” is missing or not working. This enzyme breaks down certain fats from the food we eat into energy. It also breaks down fat already stored in the body.

What Symptoms or Problems Occur with VLCAD?
[Symptoms are something out of the ordinary that a parent notices.]
There are three forms of VLCAD — infant, childhood, and adult. Symptoms can be mild or serious. Infant and childhood types of VLCAD may cause periods of illness called Metabolic Crises, or low blood sugar. Some of the first signs of a Metabolic Crisis are:
- Too much sleepiness
- Behavior changes (such as crying for no reason)
- Irritable mood
- Poor appetite

If a Metabolic Crisis is not treated, a child with VLCAD can develop:
- Breathing problems and seizures
- Coma, sometimes leading to death

Other problems include enlarged liver, enlarged heart, and muscle problems.

What is the Treatment for VLCAD?
The following treatments are often used for children with VLCAD:

1. Do not go a long time without food – Babies and young children with VLCAD should eat often to avoid low blood sugar or a Metabolic Crisis. They shouldn’t go without food for more than 4 to 6 hours. Some babies may need to eat even more often. Children with VLCAD should have a starchy snack (such as bread, cereal, rice) before bed and another during the night. They need another snack first thing in the morning. Raw cornstarch mixed with water, milk, or other drink is a good source of long-lasting energy. Your dietitian can give you ideas for good night-time snacks.

2. Diet – Sometimes your child will need to eat a diet low in fat (lean meat and low-fat dairy foods) and high in carbohydrates (such as bread, noodles, fruits, vegetables). Your dietitian will make any needed diet changes. Dietitians know what are the correct foods to eat.

3. MCT oil and L-carnitine and other supplements – Your doctor may prescribe MCT oil. This special oil has medium chain fatty acids that can be used in small amounts for energy. Sometimes the doctor will prescribe L-carnitine. This is safe and natural and helps the body make energy.

Things to Remember
Always call your doctor when your child has any of the following:
- Poor appetite
- Too much sleepiness
- Vomiting
- Diarrhea
- An infection
- A fever
- Continued muscle pain or weakness
- Reddish-brown color to the urine

Children with VLCAD need to eat extra starchy food (such as bread, cereal, rice) and drink more fluids during any illness. When they become sick, they often need to be treated in the hospital to prevent serious health problems.
Propionic Acidemia
(PROP or PA)

What is PROP?
PROP is a type of organic acid disorder. People with PROP can’t break down certain amino acids from food.

What Causes PROP?
Enzymes help start chemical reactions in the body. PROP happens when an enzyme called “propionyl CoA carboxylase” (PCC) is missing or not working. This enzyme changes certain amino acids so the body can use them. Glycine and propionic acid build up in the blood and cause problems when PCC doesn’t work.

What Symptoms or Problems Occur with PROP?
[Symptoms are something out of the ordinary that a parent notices.]
PROP causes periods of illness called Metabolic Crises. Early symptoms of a Metabolic Crisis are:
- poor appetite
- low muscle tone (floppy muscles and joints)
- too much sleepiness or lack of energy
- vomiting

If a Metabolic Crisis isn’t treated, a child with PROP can develop:
- breathing problems
- seizures
- swelling of the brain
- stroke
- coma, sometimes leading to death

Later problems can include:
- mental retardation
- low ability to fight illnesses
- osteoporosis (weak bones)
- inflamed pancreas gland
- skin rashes
- poor growth

What is the Treatment for PROP?
The following treatments are often used for children with PROP:

1. Low-protein diet, medical foods and medical formula – The best treatment for PROP is a diet low in protein. Most of the low-protein food will be carbohydrates (such as bread, cereal, noodles, fruits, vegetables). High-protein foods that should be limited or not eaten at all include:
   - milk and milk products
   - meat and poultry
   - fish
   - eggs
   - dried beans and peas
   - nuts and peanut butter

   The doctor may prescribe a special medical formula with the right amount of protein. There are also medical foods available for people with PROP.

2. Do not go a long time without food – Some babies and young children can have a Metabolic Crisis if they don’t eat often enough. They shouldn’t go without food for more than 4 to 6 hours. Some children may need to eat even more often. Your dietitian can give you ideas for suitable snacks. Dietitians know what are the right foods to eat.

3. Medication – The doctor may prescribe L-carnitine for your child. This is safe and natural and helps the body make energy.

Things to Remember
Even minor illness such as a cold or flu can cause a Metabolic Crisis. Call your doctor right away when your child has any of the following:
- loss of appetite
- vomiting
- diarrhea
- infection or illness
- fever

Many children with PROP must be treated in the hospital during illness to avoid serious health problems.
Multiple Carboxylase Deficiency
(MCD)

**What is MCD?**
MCD is an organic acid disorder. People with MCD can’t change protein and carbohydrates from the food they eat into energy for the body.

**What Causes MCD?**
Enzymes help start chemical reactions in the body. The enzyme holocarboxylase synthetase (HCS) adds the vitamin biotin to other enzymes called “carboxylases.” These change the food we eat into energy. Harmful matter builds up in the blood and urine when the HCS enzyme doesn’t work. This can cause serious health problems.

**What Symptoms or Problems Occur with MCD?**
*Symptoms are something out of the ordinary that a parent notices.*
MCD causes periods of illness called Metabolic Crises. Some of the first symptoms are:
- poor appetite
- vomiting
- too much sleepiness or lack of energy
- irritable behavior
- low muscle tone (floppy muscles and joints)
If a Metabolic Crisis is not treated, a child with MCD can develop:
- breathing problems
- seizures
- brain swelling
- coma, sometimes leading to death

Later problems can include:
- mental retardation
- skin rash and hair loss
- unsteady movements
- rigid movement
- vision and hearing loss

**What is the Treatment for MCD?**
Biotin is usually the only treatment for MCD. Your doctor will prescribe the right amount. This will prevent symptoms in your child. It may rid your child of health problems when symptoms already exist. Early treatment prevents mental retardation and other serious medical problems. Treatment should start as soon as you know your child has MCD. Your child will need to take biotin for life.

**Things to Remember**
Babies who get treatment before they have a Metabolic Crisis usually have normal growth and development.

Some children develop learning problems even when treated. Children may also develop mental retardation. Treatment may not help the problems that already exist.
What is MMA?
MMA is an organic acid disorder. People with MMA can’t break down and use certain amino acids and fatty acids from the food they eat.

What Causes MMA?
Enzymes help start chemical reactions in the body. Special enzymes break down certain amino acids and fatty acids from the protein in food so that the body can use them. MMA happens when one of these special enzymes is missing or not working. There are a number of different kinds of MMA. Some improve with Vitamin B12 injections (Vitamin B12-responsive), and some do not (Vitamin B12 non-responsive).

What Symptoms or Problems Occur with MMA?
(MMA)

What is the Treatment for MMA?
1. Medication – Vitamin B12 shots are the main treatment for Vitamin B12-responsive MMA, caused by not enough cobalamin A & B. Vitamin B12 helps most children with the first form (A). It helps about close to half of children with second form (B). Your child's doctor may prescribe L-carnitine. This is safe and natural and helps body cells make energy. Antibiotics may help.

2. Low-protein diet, medical foods and medical formula – Foods high in protein should be limited or not eaten at all. They include:
   - milk and milk products
   - meat and poultry
   - fish
   - eggs
   - dried beans and peas
   - nuts and peanut butter

The doctor may give your child a special medical formula. A dietitian will tell you what kind of formula is best and how much to use. Dietitians know what are the right formulas and foods to eat.

Things to Remember
Even minor illness can lead to a Metabolic Crisis in children with MMA. Call your doctor right away when your child has any of the following:
   - loss of appetite
   - vomiting
   - diarrhea
   - infection or illness
   - fever

Children need extra fluids and starchy food (such as bread, rice, cereal, noodles) when they’re sick in order to prevent a Metabolic Crisis. During illness, you should limit protein and give your child starchy foods and fluids. Sick children with MMA may need to be treated in the hospital to avoid serious health problems.
Newborn Screening FACT Sheet

Phenylketonuria
(PKU)

What is PKU?
PKU is a type of amino acid disorder. People with PKU have problems breaking down an amino acid called phenylalanine from the protein in food they eat.

What Causes PKU?
Enzymes help start chemical reactions in the body. PKU happens when an enzyme called “phenylalanine hydroxylase” (PAH) is either missing or not working right. This enzyme breaks down the amino acid phenylalanine (Phe). When a child with PKU eats food containing Phe, it builds up in the blood and causes problems. Phe is found in almost every food, except pure fat and sugar. PKU is an inherited disorder where both parents carry a gene for PKU.

What Symptoms or Problems Occur with PKU?

[Symptoms are something out of the ordinary that a parent notices.]

Babies with PKU seem perfectly normal at birth. The first symptoms are usually seen around 6 months of age. Untreated infants may be late in learning to sit, crawl, and stand. They may pay less attention to things around them. A child with PKU who doesn’t get treatment will become mentally retarded.

Some of the things caused by untreated PKU include:

- mental retardation
- behavior problems (such as hitting, biting)
- hyperactivity (over-active)
- restlessness or irritable mood
- seizures
- eczema (itchy areas of skin that become flaky or hard)
- a “musty” or “mousy” body odor
- light hair and skin

What is the Treatment for PKU?
The following treatments are often used for children with PKU:

1. Medical formula with low Phe – Even though they need less Phe, children with PKU still need a certain amount of protein. A special low-Phe medical formula gives babies and children with PKU the nutrients and protein they need. It helps keep their Phe at a safe level.

2. Low-Phe food plan – The right diet is made up of foods that are very low in Phe. This means your child must not have cow’s milk, regular formula, meat, fish, eggs, or cheese. Regular flour, dried beans, nuts, and peanut butter also have Phe. They must be highly limited or not eaten at all. This diet is needed for life.

Things to Remember

Children with PKU who start treatment soon after birth usually have normal growth and intelligence. Even when treated, some children have problems with schoolwork and may need extra help.
Sickle Cell Anemia
(Hb-SS)

What is Sickle Cell Anemia?
Sickle Cell Anemia (Hb-SS-Disease) is the most common Sickle Cell Disease. It clogs blood vessels and causes severe pain. It can also cause problems such as organ and tissue damage. Other common Sickle Cell Diseases are Hb-SC-Disease and sickle thalassemias.

What Causes Sickle Cell Anemia?
Sickle Cell Anemia is an inherited blood disease. It is very serious for infants and young children. A person with this life-long disease has abnormal red blood cells. Changes in these cells cause them to be shaped like a “sickle.” These red blood cells are very weak and sometimes rigid. They can get trapped in the blood vessels and harm blood flow. This can cause sudden pain anywhere in the body. It can also damage body tissues and organs over time.

What Problems Occur with Sickle Cell Anemia?
Many young children with sickle cell anemia are rarely sick. Sometimes serious problems happen that can cause death. These problems are:
- septicemia (infection of the blood)
- acute splenic sequestration (sudden enlargement of the spleen and rapid drop in the blood count)

Other serious problems can include:
- pneumonia
- organ damage
- painful erections in men
- swelling of hands and feet
- problems during pregnancy
- chest pains and trouble breathing
- blood in urine
- fever
- stroke
- leg ulcers
- infections
- jaundice (yellow skin)
- gallstones
- anemia
- pain
- kidney failure

What is the Treatment for Sickle Cell Anemia?

Medication – Infants and young children with Sickle Cell Anemia sometimes get infections of the blood. Penicillin can prevent death from this. When given twice a day, penicillin can kill bacteria before they grow in the blood and cause very dangerous infections.

Fluids – It is important to drink plenty of fluids to keep blood vessels open.

Things to Remember
Children with sickle cell anemia can have periods of pain. This happens more in older children, but sometimes happens in babies. Usually the pain seems to be in the bone. Sometimes it is in the stomach. Such pain is not dangerous. It usually goes away after several hours or days.

Medications such as acetaminophen (such as Tylenol) or ibuprofen (such as Advil) often help the pain. Sometimes children won’t use the part of the body that hurts, even when it feels better. Don’t force your child to stand or walk. Children will be active again when they feel like it. Rest and drinking plenty of liquids can help the pain. A heating pad can also help. If a child’s pain isn’t better after taking medicine at home, he or she may need to be treated with stronger medicine in the hospital.
Maple Syrup Urine Disease (MSUD)

What is MSUD?
MSUD is a type of amino acid disorder. It is named for the sweet maple syrup smell of the urine in untreated babies. People with MSUD have problems breaking down certain amino acids from protein in food.

What Causes MSUD?
Enzymes help start chemical reactions in the body. The most common form of MSUD is caused by the lack of a group of enzymes called “branched-chain ketoacid dehydrogenase” (BCKAD). This enzyme group breaks down three different amino acids. These are called “branched chain amino acids” (BCAA). When the body can't break them down, they build up in the blood and cause problems.

What Symptoms or Problems Occur with MSUD?
[Symptoms are something out of the ordinary that parents notice.]
Symptoms start as soon as a baby is fed protein, usually right after birth. Some of the first are:
- poor appetite
- weak suck
- weight loss
- high-pitched cry
- urine that smells like maple syrup or burnt sugar

Babies with MSUD have periods of illness called Metabolic Crises. Some first symptoms are:
- too much sleepiness
- tiredness
- irritable mood
- vomiting

Brain damage can occur if untreated. This can cause mental retardation. Some babies become blind. Most babies die within a few months if not treated. There are milder forms that may cause mental retardation.

What is the Treatment for MSUD?
The following treatments are often used for children with MSUD:

1. Medical Formula – The doctor may prescribe a special medical formula with the right amount of protein. This will help keep your child's BCAA levels in a safe range.

2. Diet low in BCCAs – The right diet is made up of foods that are very low in the BCAAs. This means your child will need to not eat foods such as cow's milk, regular formula, meat, fish, cheese, and eggs. Regular flour, dried beans, nuts, and peanut butter also must be highly limited or not eaten. Many vegetables and fruits can be eaten in the right amounts. Your child should use this diet for life.

3. Checking BCAA levels – Your child will have regular blood tests to measure amino acid levels. The diet and formula may need to be changed based on blood test results.

Things to Remember
For children with MSUD, even minor illness such as a cold or flu can cause a Metabolic Crisis. Call your doctor right away when your child has any of the following:
- poor appetite
- low energy or too much sleepiness
- vomiting
- an infection or illness
- a fever
- behavior or personality changes (such as crying too much)
- difficulty walking or balance problems

Children with MSUD need to eat more starchy foods (such as rice, cereal, bread) and drink more fluids during any illness or they could have a Metabolic Crisis. Children who are sick may not want to eat. If they can't eat, or if they show signs of a Metabolic Crisis, they may need to be treated in the hospital.
Tyrosinemia
(TYR1)

What is TYR1?
TYR1 is an amino acid disorder. People with TYR1 can’t break down the amino acid tyrosine from the food they eat. This causes severe liver disease and other health problems if not treated.

What Causes TYR1?
Enzymes help start chemical reactions in the body. TYR1 happens when the enzyme fumarylacetoacetase (FAH) is missing or not working. When FAH doesn’t work, it can’t break down tyrosine. Tyrosine and other harmful things then collect in the blood. One of these is succinylacetone. This causes serious liver and kidney damage. It can also cause periods of weakness or pain.

What Symptoms or Problems Occur with TYR1?
(Symptoms are something out of the ordinary that a parent notices.)

Babies usually show symptoms in the first few months of life. Some of the first signs are:
- diarrhea and bloody stools
- vomiting
- poor weight gain
- too much sleepiness
- irritable mood
- “cabbage-like” odor to the skin or urine

Liver problems are common. They can cause:
- enlarged liver
- yellow skin
- bleeding and bruising easily
- swollen legs and stomach

Kidney problems also happen and can cause:
- rickets, a bone thinning condition
- delays in walking

Babies with severe liver and kidney problems usually die without quick treatment.

What is the Treatment for TYR1?
Doctors use these treatments for children with TYR1:

1. Medication – The doctor may prescribe Nitisinone (Orfadin®). This prevents liver and kidney damage. It also stops the weakness and pain. This medicine may lessen the risk of liver cancer. Your child should take Nitisinone right away.

2. Medical Formula – The doctor may prescribe a special formula with the right amount of amino acids. He or she will tell you what kind is best and how much to use.

3. Low-tyrosine diet: Most children with TYR1 need to limit foods such as cow’s milk and regular formula. Your doctor and dietitian will decide on the best food plan for your child. Dietitians know what are the right formulas and foods to eat.

4. Blood, urine, and other tests – Regular blood and urine tests will show if your child’s diet or medicine needs to be changed.

5. Liver transplantation – A liver transplant is one way to prevent liver cancer.

Things to Remember
Early treatment can prevent severe symptoms of liver and kidney problems and weakness and pain. Children who are treated early usually have normal growth and intelligence.

If treatment isn’t started right away, children may have liver or kidney damage. Rickets might be a problem and need treatment. Delays in growth and development can also happen.
Newborn Screening FACT Sheet

Trifunctional Protein Deficiency
(TFP)

What is TFP?
TFP is a type of fatty acid oxidation disorder. People with TFP deficiency can’t break down fat into energy for the body.

What Causes TFP?
Enzymes help start chemical reactions in the body. TFP happens when a group of enzymes called “trifunctional protein” is missing or not working. TFP breaks down certain fats from the food we eat into energy. It also breaks down fat already stored in the body.

What Symptoms or Problems Occur with TFP?

[Symptoms are something out of the ordinary that a parent notices.]

Babies and children with early and childhood TFP have periods of illness called Metabolic Crises. Some of the first symptoms of a Metabolic Crisis are:

- too much sleepiness
- behavior changes (such as crying for no reason)
- irritable mood
- muscle weakness
- poor appetite

If a Metabolic Crisis is not treated, a child with TFP can develop:

- breathing problems
- seizures
- coma, sometimes leading to death

What is the Treatment for TFP?
The following treatments are often used for children with TFP deficiency:

1. **Do not go a long time without food** – Babies and young children with TFP need to eat often to avoid low blood sugar and Metabolic Crises. They should not go without food for more than 4 to 6 hours. Some babies need to eat even more often. It is important that babies be fed during the night. Your dietitian can give you ideas for good night-time snacks. Dietitians know what are the right foods to eat.

2. **Diet** – Sometimes your child needs a diet low in fat (such as lean meat and low-fat dairy foods) and high in carbohydrates (such as bread, noodles, fruits, vegetables). People with TFP cannot use certain building blocks of fat called “long chain fatty acids.” A dietitian can make a food plan low in these fats.

3. **MCT oil and L-carnitine** – MCT oil is often used for people with TFP. This special oil can be used in small amounts for energy. Sometimes the doctor will prescribe L-carnitine. This is safe and natural and helps body cells make energy. It also helps the body get rid of harmful wastes.

Do not use any medication without checking with your doctor.

Things to Remember
Always call your doctor when your child has any of the following:

- poor appetite
- low energy or too much sleepiness
- vomiting
- diarrhea
- an infection
- a fever
- continued muscle pain or weakness
- reddish-brown color to the urine
Sickle Beta Plus Thalassemia (Sβ+-Thalassemia)

What is Sβ+-Thalassemia?
Sickle Beta Plus Thalassemia (Sβ+-thalassemia) is a “mild” form of sickle cell disease. Your child’s red blood cells have only a small amount of the normal hemoglobin called hemoglobin A. They also have abnormal hemoglobin called hemoglobin S (sickle hemoglobin). The red blood cells have another problem called beta plus thalassemia. This causes red blood cells to be small and pale.

What Causes Sβ+-Thalassemia?
Sβ+-thalassemia is an inherited condition. Hemoglobin S comes from one parent. Beta plus thalassemia comes from the other parent. Instead of appearing round (like donuts), your child’s red blood cells are somewhat small, pale and misshapen.

What Symptoms or Problems Occur with Sβ+-Thalassemia?
(Symptoms are something out of the ordinary that a parent notices.)

Periods of pain can happen with Sβ+-thalassemia. The red blood cells are rigid and stiff. Sometimes they “clog up” the small blood vessels in the bones and other parts of the body. This can cause pain because not enough oxygen can get into the bones and other parts of the body. The pain usually happens in the back, stomach, arms, and legs. There may be swelling. The pain can last for a few hours or up to a week or more. The amount of pain varies. Sometimes children with Sβ+-thalassemia have a slightly enlarged spleen (an organ located on the upper left side of the stomach area). A bigger spleen usually doesn’t cause any problems.

What is the Treatment for Sβ+-Thalassemia?
Medication – There are medicines to help ease the pain. Taking medication such as acetaminophen (Tylenol) or ibuprofen (Advil) usually helps the pain.

Fluids – Your child has more risk of getting infections, especially pneumonia. The abnormal red blood cells “clog up” the lungs. This increases the risk of infection there. Your child will need to drink plenty of fluids to keep blood vessels open.

Things to Remember
Children with Sβ+-thalassemia can have normal lives and life spans. Don’t think of your child as “sick.” You should treat him or her normally. Your child will need to see the doctor for regular checkups and vaccinations. He or she will also need to make several visits a year to see a hematologist (a doctor who is a blood specialist).
Sickle Beta Zero Thalassemia
(S^0β-Thalassemia)

What is S^0β-Thalassemia?
Hemoglobin in the red blood cells carries oxygen to all parts of the body. With Sickle Beta Zero Thalassemia (S^0β-Thalassemia), the red blood cells have abnormal hemoglobin. This is called hemoglobin S (sickle hemoglobin). Also, there is a problem called thalassemia. This causes red blood cells to be small and pale.

What Causes S^0β-Thalassemia?
S^0β-Thalassemia is an inherited disease. Hemoglobin S is inherited from one parent. Thalassemia is inherited from the other parent. In S^0β-Thalassemia the red blood cells are not round. They are misshapen. Some may have a sickle (or banana) shape.

What Symptoms or Problems Occur with S^0β-Thalassemia?
(Symptoms are something out of the ordinary that a parent notices.)
The symptoms of S^0β-Thalassemia depend on the type and seriousness of the disease. Problems happen when different parts of the body don’t get enough oxygen. This occurs when there is low hemoglobin and not enough red blood cells in the blood (anemia).

Problems include:
- feeling tired and weak
- slightly enlarged spleen (an organ located on the upper left side of the stomach area)
- mild anemia
- periods of pain
- increased risk for infections

What is the Treatment for S^0β-Thalassemia?
Medications – Your child has more risk of getting infections. These can be infection in the brain, pneumonia, blood poisoning, and bone infection. Your child’s doctor will probably give him or her penicillin to help prevent infections. It is important that your child take the penicillin as directed. Any fever of 101°F (38.5°C) is serious. Take your child to the doctor or clinic IMMEDIATELY if this happens.

Periods of pain can happen with S^0β-Thalassemia. The red blood cells are rigid and stiff. Sometimes they “clog up” the small blood vessels in the bones and other parts of the body. This can cause pain because not enough oxygen can get into the bones and other parts of the body. The pain usually happens in the back, stomach, arms, and legs. There may be swelling. Pain can last for a few hours or up to a week or more. The amount of pain varies. Medicines help ease the pain.

Sometimes children with S^0β-Thalassemia have a slightly enlarged spleen. A bigger spleen usually doesn’t cause any problems.

Things to Remember
Children with S^0β-Thalassemia can have normal lives and life spans. Don’t think of your child as “sick.” You should treat him or her normally. Your child will need to see the doctor for regular checkups and vaccinations. He or she will also need to make several visits a year to see a hematologist (a doctor who is a blood specialist).
What is Hb S/C?
Hemoglobin Sickle C Disease (Hb S/C) is a “mild” form of sickle cell anemia. The red blood cells (RBCs) of a child with Hb S/C disease have two kinds of abnormal hemoglobin. They are called hemoglobin S and hemoglobin C.

What Causes Hb S/C?
The hemoglobin in the RBCs carries oxygen to all parts of the body. Regular RBCs are round like a donut. The RBCs in a child with Hb S/C disease are misshapen. Some even look like the sickle-shaped cells found in sickle cell anemia. Others are folded or football-shaped.

What Symptoms or Problems Occur with Hb S/C?
(Symptoms are something out of the ordinary that a parent notices.)

Anemia – Your child will always have a slightly low red blood count. This is called anemia. This mild anemia is usually no problem, but sometimes causes tiredness and/or weakness.

Pain – The red blood cells of Hb S/C disease are rigid and stiff. Sometimes they “clog up” the small vessels in the bones and other parts of the body. This can cause pain because enough oxygen cannot get into the bones. The pain usually happens in the arms, legs, stomach, and/or back. It can last for hours, for days, or up to a week. It can vary from mild to moderate to severe. The location, length, and amount of pain can vary. How often this happens also varies. Some children with Hb S/C disease have no periods of pain at all, but most will have a few each year.

Pneumonia and Other Infections – A child with Hb S/C disease has a higher chance of getting certain infections, especially pneumonia. The abnormal RBCs can “clog up” in the lungs and cause infection. This is called “chest syndrome.” Problems to watch for include fever, fast breathing, trouble breathing, retractions (ribs “suck in” when breathing), very congested cough, and chest pain. Your child should see a doctor immediately if these problems happen.

Sickle C Disease
(Hb S/C)

Spleen – The spleen is a small organ located on the upper left side of the stomach area, up under the rib cage. It helps fight infection in the body. Children with Hb S/C disease may have an enlarged (big) spleen. This doesn’t happen until they are about five years or older. A big spleen usually doesn’t cause any problems. Sometimes teen-agers and/or adults can have pain near the spleen and a drop in the red blood count. This is called a “spleen crisis.”

Eyes – Older children (over age ten) and adults with Hb S/C disease may develop damage to the retina in the back of the eye. This can cause blindness if not treated early. Regular eye checkups by a medical eye doctor (ophthalmologist) are needed to diagnose and treat this problem.

What is the Treatment for Hb S/C?
Medication – The infant or young child with Hb S/C disease is more likely to have infection of the blood (septicemia). Scientific studies show that penicillin can prevent death from septicemia. When taken every 12 hours, penicillin can kill bacteria before they grow in the blood and cause septicemia.

Fluids – The infant or young child with Hb S/C disease needs to drink plenty of liquids to keep blood vessels open.

Things to Remember
Children with Hb S/C disease can experience periods of pain. Medications for pain, such as acetaminophen (Tylenol) or ibuprofen (Advil) often help the pain. Sometimes children won’t use the part of the body that hurts, even when it feels better. Don’t force your child to stand or walk. Children will be active again when they feel like it. Rest and drinking plenty of liquid can help the pain. A heating pad can also help. If a child’s pain isn’t better after taking medicine at home, he or she may need to be treated with a stronger medicine in the hospital.