Fact Sheets

Information for **PARENTS** about the disorders included in the Expanded Newborn Screening Panel

Table of Contents

3– METHYLCROTNYL CoA CARBOXYLASE DEFICIENCY [3-MCC]

What is 3-MCC?

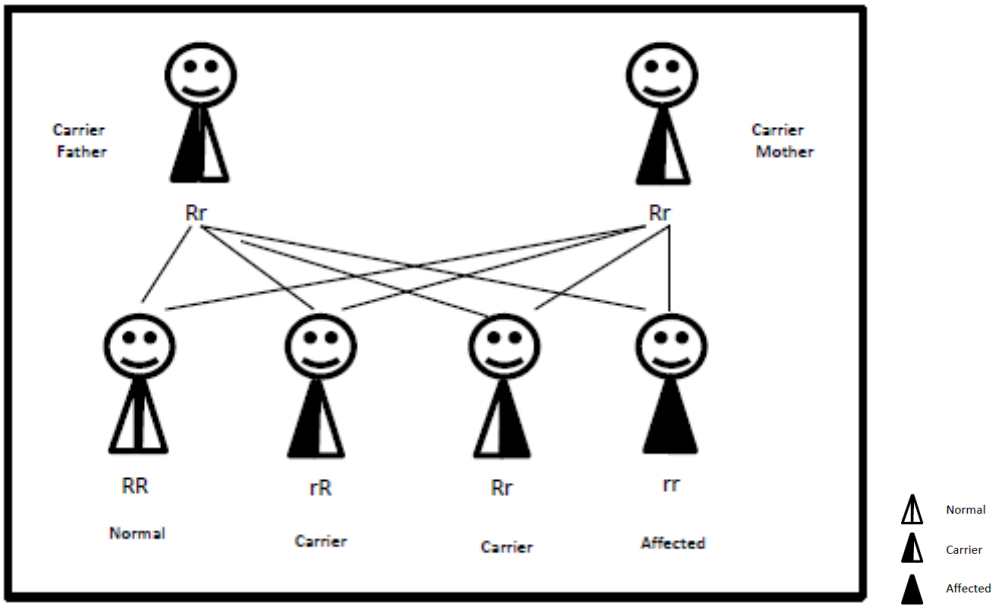
3-MCC is a condition that is due to a deficiency in an enzyme or chemical scissors called 3-methylcrotonyl CoA carboxylase which is needed to breakdown an amino acid called leucine. Amino acids are the building blocks of protein. Children with this condition will look normal at birth. Untreated children may remain without symptoms while others may have seizures, drowsiness, low muscle tone, poor appetite and failure to thrive. This disorder can be detected through newborn screening. However, neonates who test positive for this condition in expanded newborn screening do not actually have the condition but instead reflect the increased levels of the metabolites of their mothers. Thus further investigation of the mother should be done as well.

What causes 3-MCC?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors, children with this condition cannot effectively breakdown the amino acid leucine. The accumulation of leucine causes the signs and symptoms of this condition.

3-MCC is an inherited condition. The gene for the 3-methylcrotonyl CoA carboxylase enzyme is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with 3-MCC have one working and one non-working gene coding for a particular enzyme needed in the breakdown of fat. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have a 3-MCC. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of 3-MCC?

Children with this condition will look normal at birth. Untreated children may remain without symptoms while others may have seizures, drowsiness, low muscle tone, failure to thrive and poor appetite.

What is the treatment of 3-MCCC ?

The main treatment of a 3-MCC is through a low protein diet. If the child is well or does not have any illness, he/she should eat a low protein diet. Glycine and/or Carnitine, a medication that helps boost energy, is also given.

What should I do when my baby is unwell?

Children with 3-MCC may have a **metabolic crisis** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drank well or during stressful events (such as surgery and severe infection). Your child may present with lethargy, seizures or convulsions, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

BETA-KETOTHIOLASE DEFICIENCY

What is Beta-ketothiolase Deficiency?

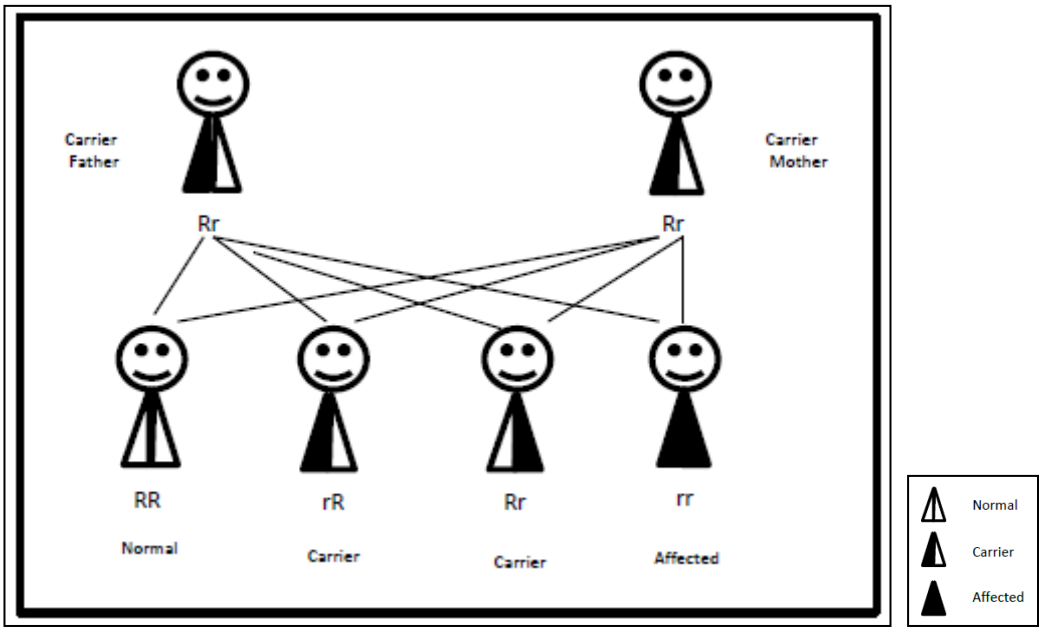
Beta-ketothiolase Deficiency is due to a deficiency in the mitochondrial acetoacetyl-CoA enzyme or chemical scissor that breaks down amino acids (or the building blocks of protein) in the body. Children with this condition cannot properly breakdown protein in their body or from the food they eat. Children born with this condition appear normal at birth but may present with severe vomiting and diarrhea, low blood sugar, seizures and coma. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition such as low blood sugar, increased sleepiness, acidosis, coma and death.

What causes Beta-Ketothiolase Deficiency?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissor, children with this condition cannot effectively utilize protein and its building block called amino acids (specifically isoleucine) from their body or from the food they eat.

Beta-ketothiolase is an inherited condition. The gene for the mitochondrial acetoacetyl-CoA enzyme is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with Beta-Ketothiolase deficiency have one working and one non-working gene coding for a particular enzyme needed in the breakdown of fat. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have a Beta-Ketothiolase Deficiecy. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of Beta-Ketothiolase Deficiency?

Children born with this condition appear normal at birth but if treatment is not given early, they can present with low blood sugar which can lead to seizures, coma and death. They may also have acidosis, vomiting and diarrhea.

What is the treatment of Beta-Ketothiolase Deficiency?

The main treatment of Beta-Ketothiolase Deficiency is through a low protein diet. If the child is well or does not have any illness, he/she should eat regular meals and avoid fasting more than 6 hours.

Carnitine, a supplemental medication essential for muscle energy production has been found to be of benefit for some patients.

What should I do when my baby is unwell or has an illness (like respiratory or gastrointestinal infection)?

Children with Beta-Ketothiolase Deficiency may have a "**metabolic crisis"** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drank well or during stressful events (such as surgery and severe infection). Your child may present with lethargy, seizures or convulsions, irritability and vomiting and acidosis. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

BIOTINIDASE DEFICIENCY

What is Biotinidase Deficiency?

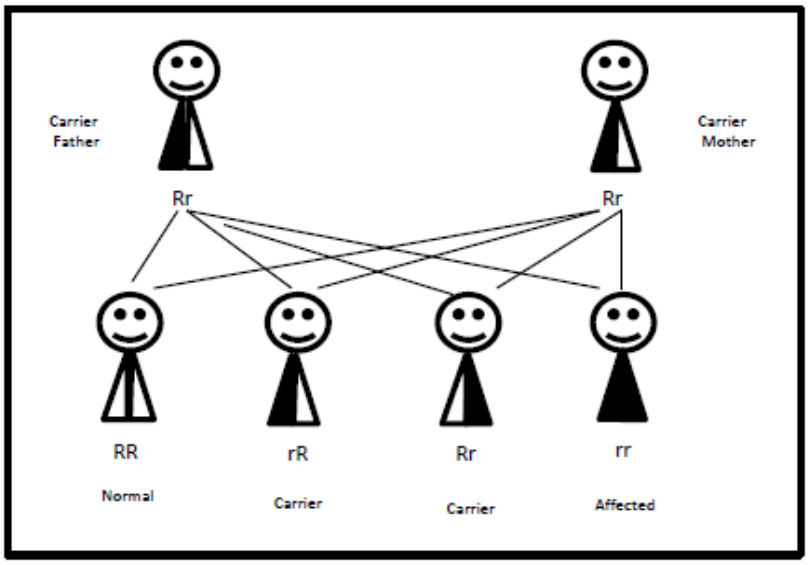
Biotinidase Deficiency is due to a deficiency of an enzyme (or chemical scissors) called biotinidase. Biotinidase helps in the production of biotin. Biotin is a vitamin that is required by carboxylases which are other enzymes needed to change the food we eat into energy. Untreated children born with this condition may present with drowsiness, poor appetite, poor weight gain, vomiting and a skin rash. They may have mental retardation and brain damage as a complication. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition.

What causes Biotinidase Deficiency?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors, children with this condition cannot effectively breakdown certain carbohydrates and fats which in turn are needed to breakdown protein.

The gene for the biotinidase enzyme is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with biotinidase deficiency have one working and one non-working gene coding for this enzyme. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have biotinidase deficiency. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of Biotinidase Deficiency?

Untreated children born with this condition may present with drowsiness, poor appetite, poor weight gain, vomiting and a skin rash. They may have mental retardation and brain damage as a complication.

What is the treatment of Biotinidase Deficiency?

The main treatment of biotinidase deficiency is through dietary supplementation with biotin.

CARNITINE PALMITOYL TRANSFERASE DEFICIENCY TYPE 1 [CPT1 deficiency]

What is Carnitine Palmitoyl Transferase Deficiency Type I?

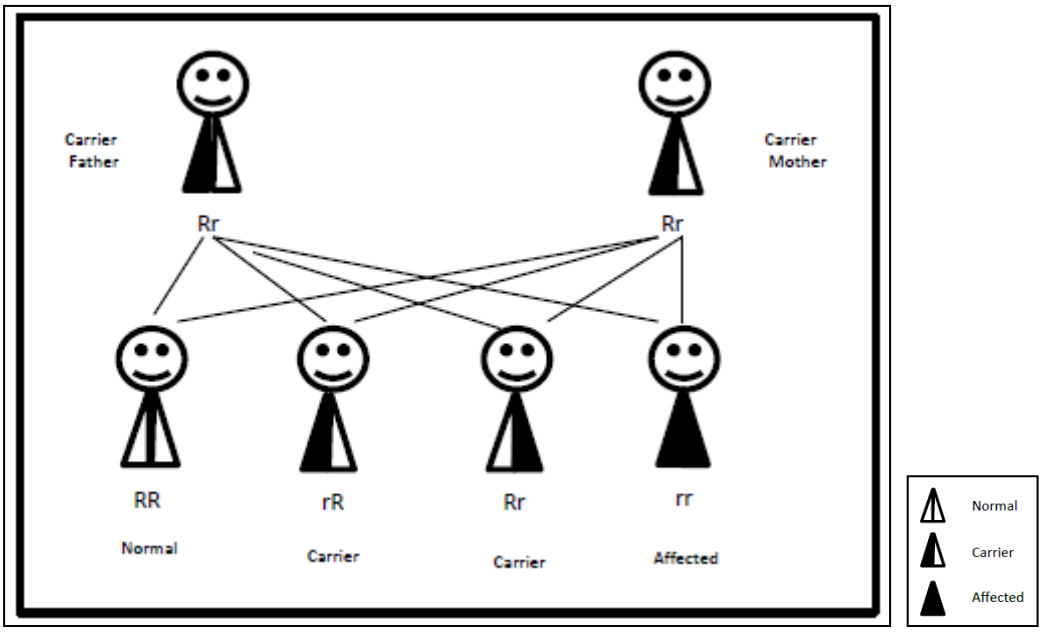
Carnitine Palmitoyl Transferase Type I (CPT1) Deficiency is due to a deficiency in an enzyme or chemical scissor called Carnitine Palmitoyl Transferase Type I that breaks down fats in the body. Children with this condition cannot properly breakdown fat in their body or from the food they eat. Children born with this condition appear normal at birth but if treatment is not given early, they may present with low blood sugar and lead to seizures, coma and death. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition such as low blood sugar, liver disease, increased sleepiness and muscle weakness.

What Causes CPT 1 Deficiency?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissor, children with this condition cannot effectively utilize fat from their body or from the food they eat.

CPT1 Deficiency is an inherited condition. The gene for the different enzymes is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with CPT1 Deficiency have one working and one non-working gene coding for a particular enzyme needed in the breakdown of fat. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have CPT1 Deficiency. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of CPT1 Deficiency?

Children born with this condition appear normal at birth but if treatment is not given early they can present with low blood sugar which can lead to seizures, coma and death. Muscle weakness and liver disease may be among the complications.

What is the treatment of CPT1 Deficiency?

The main treatment of CPT1 Deficiency is through a low fat diet. Children may be given a special milk formula called MCT (medium chain triglycerides). If the child is well or does not have any illness, he/she should eat regular meals and avoid fasting more than 6 hours.

What should I do when my baby is unwell or has an illness (like respiratory or gastrointestinal infection)?

Children with CPT1 may have a "**metabolic crisis"** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drank well or during stressful events (such as surgery and severe infection). Your child may present with lethargy, seizures or convulsions, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

CARNITINE PALMITOYL TRANSFERASE DEFICIENCY TYPE II [CPT2 deficiency]

What is CPT2 deficiency?

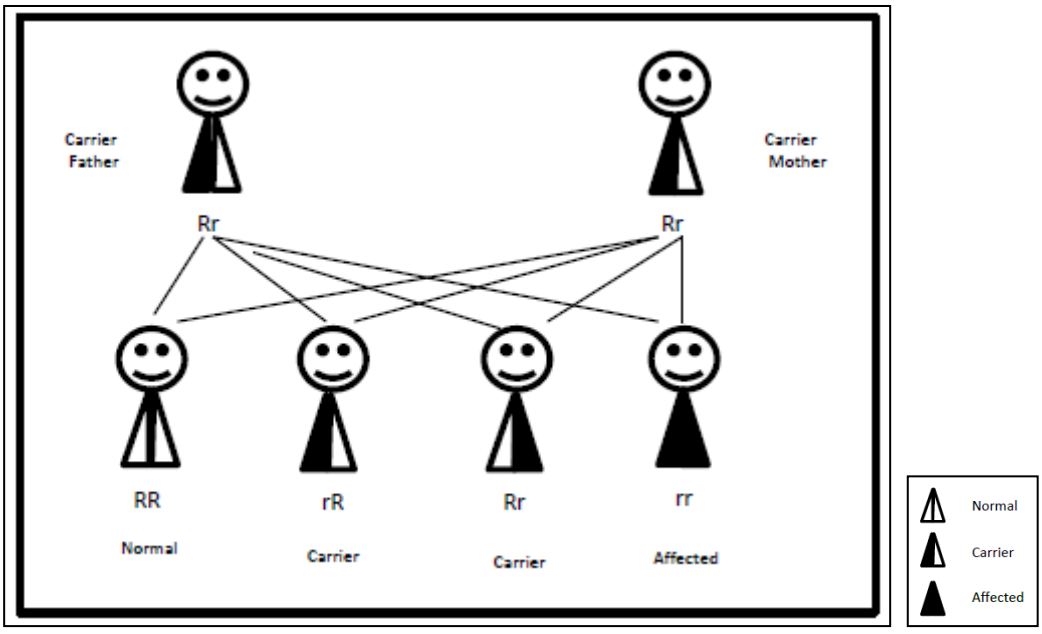
Carnitine Palmitoyl Transferase Type II (CPT2) Deficiency is due to a deficiency in an enzyme or chemical scissor called Carnitine Palmitoyl Transferase Type II that breaks down fats in the body. Children with this condition cannot properly breakdown fat in their body or from the food they eat. Children born with this condition appear normal at birth but if treatment is not given early, they may present with low blood sugar and lead to seizures, coma and death. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition such as low blood sugar, increased sleepiness, enlarged heart, liver disease and muscle weakness.

What Causes CPT 2 Deficiency?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissor, children with this condition cannot effectively utilize fat from their body or from the food they eat.

CPT2 Deficiency is an inherited condition. The gene for the different enzymes is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with CPT2 Deficiency have one working and one non-working gene coding for a particular enzyme needed in the breakdown of fat. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have CPT2 Deficiency. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of CPT2 Deficiency?

Children born with this condition appear normal at birth but if treatment is not given early they can present with low blood sugar which can lead to seizures, coma and death. Muscle weakness, liver disease and an enlarged heart may be among the complications.

What is the treatment of CPT2 Deficiency?

The main treatment of CPT2 Deficiency is through a low fat diet. Children may be given a special milk formula called MCT (medium chain triglycerides). If the child is well or does not have any illness, he/she should eat regular meals and avoid fasting more than 6 hours.

Carnitine, a natural substance that helps the body make energy, **may** also be given as supplementation based on your doctor's advice.

What should I do when my baby is unwell or has an illness (like respiratory or gastrointestinal infection)?

Children with CPT2 may have a "**metabolic crisis"** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drank well or during stressful events (such as surgery and severe infection). Your child may present with lethargy, seizures or convulsions, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

CONGENITAL ADRENAL HYPERPLASIA

What is Congenital Adrenal Hyperplasia?

CAH is an inherited condition that affects the adrenal glands and causes a number of specific health issues.

'Congenital' means the condition is present at birth. The adrenal glands are cone-shaped organs that sit on top of each kidney. They make a number of hormones necessary for healthy body function. Hyperplasia means 'overly large'. In people with CAH, the adrenal glands cannot make enough of a hormone called cortisol. As they start working harder in attempts to make more cortisol they increase in size, resulting in hyperplasia.

Babies with CAH are born with a number of physical changes. Their adrenal glands are often larger than normal, even at birth. Girls with CAH may be born with external sex organs that appear more masculine than they should. If not treated, both boys and girls will develop early sexual characteristics, well before normal puberty should begin.

What causes CAH?

Normally, the adrenal glands make a number of different hormones, including cortisol, aldosterone and androgens. Hormones are chemicals that send messages to other organs or tissues of the body, telling them to do specific things.

CAH occurs when a particular enzyme called 21-hydroxylase (21-OH) is missing or not working correctly. The job of this enzyme is to help make cortisol and aldosterone in the adrenal glands so they can be released when the body needs them.

One of the main jobs of cortisol is to keep the amount of glucose, the sugar used for energy by the body's cells, at a normal level. Cortisol also helps protect the body during times of physical or emotional stress such as surgery, injury or illness. It helps to regulate the immune response and inflammation so our bodies can deal with infection or illness.

Another hormone made by the adrenal glands is aldosterone. This hormone is released into the blood when the blood pressure drops too low. It tells the kidneys to pull salt and water out of urine and put it back into the blood. This raises blood pressure back to normal and prevents the body from losing too much fluid. Babies with a form of CAH called "salt-wasting" do not make enough aldosterone and they lose too much salt and water in their urine. They become dehydrated and their blood pressure drops too low. This can be life-threatening if not treated quickly.

The other hormones made by the adrenal glands are called androgens. These are male-like sex hormones. The adrenal glands also make a small amount of female hormones.

Most people with CAH make too much of the androgen hormones and not enough cortisol or aldosterone. Having too much of the androgen hormones in the blood causes female babies to develop masculine changes to their genitals. And, high levels of androgens lead to early sexual development, well before the normal age of puberty, in both boys and girls.

If CAH is not treated, what problems occur?

The effects of CAH can vary greatly from person to person. There are a number of different forms of CAH which are described below.

Most babies found to have CAH during newborn screening have 'classic CAH'. One type of classic CAH is called 'salt-wasting' which is a serious condition needing immediate treatment. The other type of classic CAH is called 'simple virilizing'. Children with this type do not have immediate risks to their health but still need treatment.

A small number of children are found through newborn screening to have milder or 'nonclassic CAH' which often causes fewer health problems. The symptoms of nonclassic CAH are quite variable from person to person.

Classic CAH – "Salt-wasting form"

About 75% of babies with classic CAH have the 'salt-wasting' form. Salt-wasting CAH occurs when the adrenal glands make lower amounts of both cortisol and aldosterone and too much androgen. Babies who do not make enough aldosterone will start losing too much water and salt in their urine. This can quickly cause dehydration and very low blood pressure. This can be life-threatening if not treated right away.

Infants with salt-wasting CAH usually show some of the following features within the first few weeks of life:

- Poor feeding
- Listlessness and drowsiness
- Vomiting
- Diarrhea
- Dehydration
- Weight loss
- Low blood pressure
- Low blood salt (low blood sodium level)
- Too much acid in the blood, called metabolic acidosis

If not treated, severe dehydration leads to shock, a serious situation in which not enough blood is getting to the brain and other organs. In babies with salt-wasting CAH, this is also called an "adrenal crisis". The signs of an adrenal crisis include:

- Confusion
- Irritability
- Rapid heart rate
- Coma

Periods of adrenal crisis due to too little aldosterone can occur as early as one week to one month of age. If a child in shock is not treated, there is a risk of death.

Even when carefully treated, children with salt-wasting CAH are still at risk for adrenal crises when they become ill or are under stress. The body needs more than the usual amount of adrenal hormones during illness, injury or stress. This means a child with CAH must be given more medication during these times to prevent an adrenal crisis.

All babies with salt-wasting CAH have the other features of classic CAH listed below. Girls with salt-wasting CAH usually have more male-like changes to their genitals than girls with simple virilizing CAH.

Classic CAH – Simple virilizing form

About 25% of babies with CAH have the simple virilizing form. The adrenal glands make enough aldosterone but not enough cortisol; they also make too much androgen.

Classic CAH starts its effects before birth. Excess androgen hormones are made by the fetus. This causes the genitals of female fetuses to develop male-like features. Baby girls born with classic CAH often have an enlarged clitoris. In some girls this is not very noticeable, but in others it may look like a small penis. Baby girls may also have labia which are fused together, may be wrinkled and may look more like a male scrotum. Some baby girls have fewer genital changes than others. The high level of androgen hormones does not affect the uterus and ovaries, which develop normally.

Girls who are not treated may develop other male-like traits and behaviors as they grow. Some of these changes may include:

- Deep, husky voice
- Excess hair on the face and body
- Lack of menstrual periods or scanty or irregular periods
- Early puberty changes such as hair in the armpits and pubic area
- Severe acne
- Male-pattern baldness (loss of hair near the temples)

Boys who are not treated may have some of the following traits:

- Muscle growth at an early age
- Pubic hair and underarm hair during childhood
- Enlargement of the penis during childhood
- Early deepening of the voice
- Early beard
- Smaller than normal testicles
- Severe acne

Sometimes the changes of early puberty happen in boys and girls as young as two to four years old. Both boys and girls may have rapid growth during childhood but end up being short as adults. Excess androgen hormones in childhood cause the rapid growth. The androgens also cause shorter adult height by closing the growth plates too soon.

Some untreated adults also have problems with infertility and may have difficulty achieving pregnancy.

Children with simple virilizing CAH are at risk for adrenal crises, though typically less severe than seen in children with the salt-wasting form. Acute illness or stress increases the body's need for cortisol. If children with CAH do not receive increased amounts of medication during illness or stress, they are at risk for health problems.

Nonclassic CAH / Late-onset CAH

Nonclassic CAH, also called 'late-onset', usually causes milder effects than classic CAH. However, symptoms can be quite variable from person to person. Many people with nonclassic CAH often start showing signs during childhood, adolescence, or early adulthood. Some people never develop symptoms. Newborn screening can detect some, though not all, babies with the nonclassic form of CAH.

Babies with nonclassic CAH are usually healthy at birth and their genitals are normal in appearance. They do not have salt-wasting and are not at risk for adrenal crises.

Children and adults with nonclassic CAH have adrenal glands that make near-normal amounts of cortisol and normal amounts of aldosterone. However, they have too much 17-OH progesterone (17-OHP), a chemical used to make cortisol, in their blood. They also may make too much of the androgen hormones.

Some of the traits that are sometimes seen in both males and females with non-classic CAH include:

- Rapid growth in childhood and early teens with short adult height
- Severe acne

Early puberty with development of pubic hair, underarm hair and body odor during childhood
Excess hair on the face and other parts of the body
Male-pattern baldness (hair loss near the temples)

Girls and women may have:
Male-like changes in physical appearance and behavior
Irregular menstrual periods or early-onset of periods
Infertility
Polycystic ovary syndrome

Boys may have:
Early beard growth
Enlarged penis
Small testicles

What is the treatment of CAH?

Your baby's primary doctor may work with a pediatric endocrinologist to provide medical care to your child. It is important for babies with classic CAH to be diagnosed as quickly as possible. This allows treatment to begin soon after birth which helps reduce the effects of CAH.

The main treatment for classic CAH is a drug called 'hydrocortisone' (also called 'cortisone'), taken in pill form. This medication replaces the cortisol that your baby cannot make on his or her own. It must be taken daily throughout life to prevent effects of CAH. Cortisone is sometimes given in other drug form, such as prednisone or dexamethasone.

Treatment for Classic CAH – both simple virilizing and salt-wasting forms:

Cortisone medication

The main treatment is to replace the amount of cortisol not being made by the adrenal glands. Hydrocortisone, a synthetic form of cortisol, is given by mouth in pill form. This treatment lessens the amount of androgens, prevents early puberty, and allows for more typical growth and development. Your doctor will follow your child's growth, pubertal development, blood pressure, and hormone levels throughout childhood. The level of medication needed to control symptoms will be adjusted as needed throughout your child's life.

It is important to always follow your doctor's orders on how much cortisone to give your child. Too much cortisone can cause temporary symptoms of Cushing syndrome so the dose must be carefully balanced to your child's height, weight and activity level. Signs of Cushing syndrome include: stretch marks on the skin, rounded face, weight gain, high blood pressure, and bone loss.

In addition, your doctor will give you instructions for increasing the dose of hydrocortisone during an acute illness. If you have questions about dosing, call your doctor. The body needs more cortisol during illness, injury or times of stress. Therefore, the cortisone dosage must be increased by your doctor when your child is ill, injured, or requires surgery. If your child is ill and cannot take the pills, cortisone injections may be necessary.

Hydrocortisone must be taken throughout life to prevent CAH effects. If the medication is stopped, symptoms will develop.

Surgery for girls with classic CAH

Girls who are born with an enlarged clitoris or changes to the labia have the option of surgery to change their outer genitals to a more female appearance. Some women who have CAH have not had surgery and are happy they did not. Others are glad their parents decided to give them the surgery. This is a complex decision made by the parents with guidance from their doctors. Parents who are not sure about surgery may want to talk with other families who have faced similar decisions.

If you choose corrective surgery, it can be done as early as age one to three. Surgery on the clitoris usually hides the excess tissue but leaves the clitoris itself intact. Surgery to separate the labia and to create a normal vagina is often delayed until the teenage years. Ask your doctor about the risks and benefits of surgery for these changes and the best time to do these surgeries.

Treatment to prevent short stature

Your doctor may take periodic X-rays to check your child's 'bone age'. This allows your doctor to tell whether your child is growing at too rapid a rate. It also shows whether the growth plates are still open or whether they are closing too early.

Specific medications may help increase height in children and teens that show signs of early growth failure. Certain medications lower androgen levels. If you have questions about your child's growth, talk to your doctor about the costs and benefits of these treatments.

Treatment for early puberty

Children who show changes of puberty at a young age are sometimes treated with medications that lower the amount of androgen hormones. Your doctor will talk to you about these medications should your child start showing signs of puberty during childhood.

Additional treatment for classic CAH – salt-wasting form

Children with salt-wasting CAH need to take an additional medication called Florinef. Florinef (9a-fludrohydrocortisone) is a 'salt-retaining' drug that replaces the aldosterone absent in children with salt-wasting CAH. It is given by mouth in pill form.

Some children with salt-wasting CAH need to follow a food plan that contains more salt than usual. In addition, your doctor may recommend salt tablets to prevent dehydration. It is important to follow your doctor's instructions on how much salt to feed your child. Most children on medication do not need to add extra salt to their diets.

Treatment for nonclassic CAH

Some people with nonclassic CAH do not need treatment and may go through life without symptoms. Others begin having symptoms in childhood, adolescence or young adulthood and may need medication in the form of cortisone pills. Symptoms that may signal the need for treatment include:

- Severe acne
- Excess body hair
- Irregular menstrual periods
- Lumps in the testicles
- Infertility

Children and adults with nonclassic CAH usually need less medication than children with classic CAH.

What happens when CAH is treated?

Children with CAH who start treatment soon after birth usually have normal growth and development. In most treated children, puberty occurs at the normal age, although some still have early changes. Even when treated, some adults are shorter than average.

Girls on medication usually have normal menstrual periods. Pregnancy is possible, although fertility may be lessened in some women.

Children with salt-wasting CAH who remain on treatment usually do not have further salt-wasting adrenal crises or other associated health problems.

Pregnant women with classic salt-wasting CAH should be followed carefully by an endocrinologist during pregnancy. Medications may need to be increased during pregnancy to prevent problems with fetal growth.

What causes the 21-hydroxylase enzyme to be absent or not working correctly?

Genes tell the body to make various enzymes. People with CAH have a pair of genes that do not work correctly. Because of the changes in this pair of genes, the 21-OH enzyme either does not work properly or is not made at all.

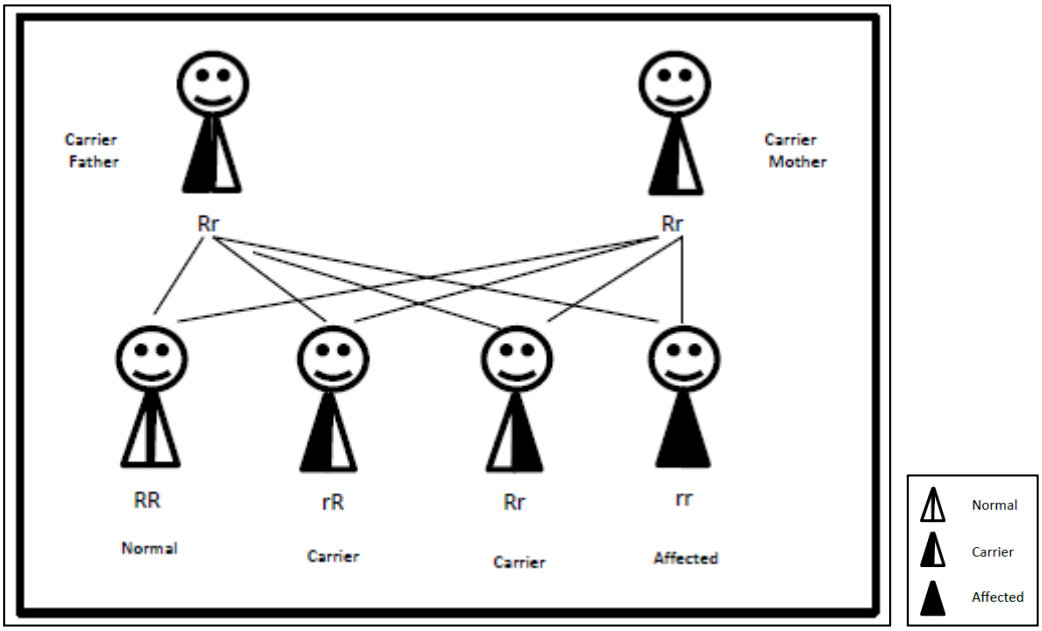
How is CAH inherited?

CAH is inherited in an autosomal recessive manner. It affects both boys and girls equally.

Everyone has a pair of genes that make the 21-OH enzyme. In children with CAH, neither of these genes works correctly. These children inherit one non-working gene for the condition from each parent.

Parents of children with CAH rarely have the condition themselves. Instead, each parent has a single non-working gene for CAH. They are called carriers. Carriers do not have CAH because the other gene of this pair is working correctly.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have CAH. There is a 50% chance for the child to be a carrier, just like the parents. And, there is a 25% chance for the child to have two working genes.



Reference:

http://newbornscreening.info/Parents/otherdisorders/CAH.html
Accessed on 7 November 2014

CONGENITAL HYPOTHYROIDISM

What is Congenital Hypothyroidism?

CH stands for "congenital hypothyroidism". Congenital means present at birth. 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.

People with hypothyroidism have specific symptoms and health issues. Babies who do not have enough thyroid hormone are often slow to grow, are sluggish, and have learning delays and other specific health problems. There are number of different causes for CH which are mentioned below.

What causes CH?

CH can be the result of a number of different underlying causes.

Missing or misplaced thyroid gland

Most babies with CH are missing their thyroid gland or have a thyroid that did not develop properly. In some cases, the thyroid gland may be smaller than usual or may not be located in the correct place.

In healthy people, the thyroid gland is located in the center of the front of the neck, near the top of the windpipe. In some children with CH, the thyroid gland may instead be under the tongue or on the side of the neck. If the thyroid gland is in the wrong place, or if it is underdeveloped, it often does not work well and makes less thyroid hormone than needed by the body. If the thyroid gland is missing, the baby cannot make any of its own thyroid hormone. A missing, underdeveloped or misplaced thyroid gland is a birth defect that happens for unknown reasons and is usually not inherited.

Hereditary causes

Less often, CH is caused by inherited changes in a gene or pair of genes. This is explained in more detail below. Children with the inherited type of CH do not make enough thyroid hormone even though their thyroid gland appears normal in size and shape. About 15% of children with CH are thought to have an inherited form.

Maternal iodine deficiency

If the mother is deficient in iodine during the pregnancy, the fetal thyroid gland may not be able to make enough thyroid hormone. The baby is then born with CH. This is a problem in some parts of the world where people do not get enough iodine in their diet. This is a very rare cause for CH in the United States because our table salt is supplemented with iodine ('iodized salt'). Other foods, in particular dairy products, contain iodine, as well.

Maternal thyroid condition and medications

In a small number of cases, CH occurs when the mother is given anti-thyroid drugs during pregnancy to treat her own thyroid problem.

If CH is not treated, what problems occur?

Most babies do not have symptoms right away because they are protected by their mother's thyroid hormone for a few weeks after birth. After about three to four weeks of age babies must rely solely on their own thyroid hormone. If they don't make enough, symptoms will show up at that time. A small number of babies with CH do show effects at birth, however.

Some babies have a yellow color to their skin or the whites of their eyes. This is called jaundice. Other signs that may occur in early infancy include:

- Low activity level - babies sleep more than usual and don't move as much
- Poor feeding and poor suck
- Fewer bowel movements or constipation
- Floppy muscle tone (hypotonia)
- Swelling around the eyes and a puffy face
- Large swollen tongue
- Cool, pale, dry skin
- Large soft spot on the skull (the fontanel) that closes late
- Large belly with protruding navel ('umbilical hernia')

If left untreated, babies may develop some or all of the following effects over time:

- 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

Children who remain untreated usually become mentally retarded and are much shorter than average. They may have spasticity and an unsteady gait. Most have speech delays and some have behavior problems.

What is the treatment of CH?

Your baby's doctor may work with a pediatric endocrinologist, a doctor with training in treating children with thyroid and other hormone problems, to care for your child.

The main treatment for CH is thyroid hormone replacement. It is safe and easy to take. If it is begun immediately after your child is diagnosed, treatment can prevent many or all of the effects of CH. If damage to the brain and nerves happens because treatment is delayed, it is usually permanent and cannot be reversed.

1. Medication

L-thyroxine is a synthetic form of thyroid hormone (but its chemical structure is identical to that produced by the normal thyroid gland). This is given in tablet form to all babies with CH. Your doctor and endocrinologist will decide how much L-

thyroxine your baby needs and how often. Your doctors will increase the amount of medication as your child grows. L-thyroxine needs to be taken on a daily basis through your child's whole life.

L-thyroxine tablets are small and can be crushed into food or dissolved into a small amount of formula, juice or other liquid. Do not dissolve them into a full bottle or glass of liquid because your baby may not finish the whole bottle and will not get the full dose of medicine. Young children can easily chew and swallow the pills. There is no approved liquid form of thyroid hormone.

It is important to give your child the correct amount of L-thyroxine. Giving your child more than he or she needs can cause body functions to speed up. Some of the signs that occur when a child takes too much L-thyroxine are:

Rapid heart rate

Diarrhea

Lack of sleep

Shakiness

Synthetic L-thyroxine is the safest form of medication to use. In the past, before synthetic forms were available, children were treated with dried thyroid hormone from pooled animal tissue. This is called 'dessicated thyroid' and is still available. Do not use dessicated thyroid as the dose of hormone is not consistent.

Soy-based formulas and iron supplements can reduce the amount of thyroid hormone your baby absorbs from the pills. Separate the time you administer your baby's thyroid medication by at least one hour from the time you feed soy formula or iron medication. Tell your doctor if you feed your baby a soy-based formula or iron supplements so the medication can be monitored and increased if necessary.

2. Monitoring

Your child will need regular visits to the doctor to check his or her weight, height, development and overall health. Your child will also likely need regular blood tests to check the level of thyroid hormone. Blood tests are usually done every one to three months until age one, and then every two to four months until age three. They can usually be done less often after age three.

3. Developmental Evaluation

Your doctor may suggest a formal evaluation of your child's development. If your child show delays in certain areas of learning or speech, extra help can be arranged. Early intervention programs are available in most states to provide services to children before they reach school age.

What happens when CH is treated?

Children with CH who start treatment soon after birth usually have normal growth and intelligence and can live typical and healthy lives. Some children, even when treated, have problems with school work and may need extra help. Some may have delayed growth compared to other children their age.

If treatment is not started until several months after birth, delays or learning problems may occur. The level of delay varies from child to child.

How is CH inherited?

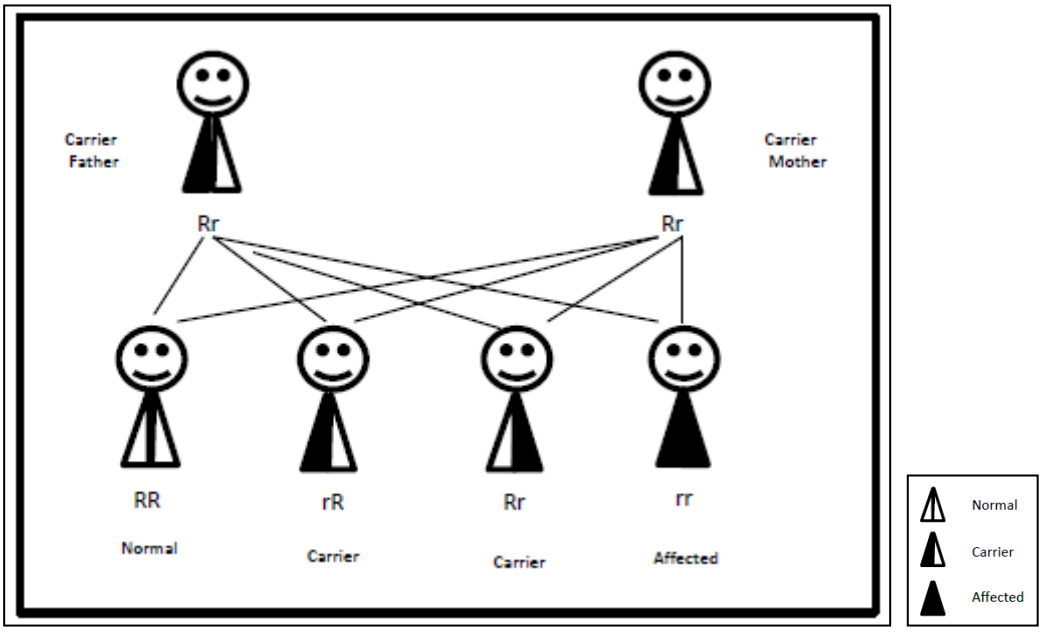
About 80 to 85% of the time, CH is caused when the thyroid gland does not develop at all, is misplaced, or is too small. Most of the time, these cases are not thought to be caused by inherited factors.

In about 15% of cases of CH, the thyroid gland appears normal but the amount of thyroid hormone made is reduced. These cases are more likely to be inherited, but not always. If an inherited form of CH is suspected, you may be referred to a genetic doctor or genetic counselor to determine whether the CH is inherited.

Most of the hereditary types of CH are inherited in an autosomal recessive manner. This type of inheritance affects both boys and girls equally. In children with autosomal recessive CH, a specific pair of genes is not working correctly and too little thyroid hormone is made. These children inherit one non-working gene for the condition from each parent. These children have a normal appearing thyroid that is in the correct place in the neck but does not make enough thyroid hormone.

Parents of children with autosomal recessive CH rarely have the condition themselves. Instead, each parent has a single non-working gene for CH. They are called carriers. Carriers do not have CH because the other gene of this pair is working correctly.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have CH. There is a 50% chance for the child to be a carrier, just like the parents. And, there is a 25% chance for the child to have two working genes.



In very rare cases, CH may be inherited in a different way, either by an X-linked recessive or autosomal dominant gene. If your child has one of these rare inherited types of CH, your genetic counselor or genetic doctor will explain how it is inherited and who else in the family may have a chance to pass on the gene for CH.

Reference:

http://newbornscreening.info/Parents/otherdisorders/CH.html
Accessed on 7 November 2014

CYSTIC FIBROSIS

What is Cystic Fibrosis?

Cystic Fibrosis is an inherited condition caused by build-up of sticky and thick mucus that can damage the body's organs. Untreated children born with this condition may have serious chronic health effects that could lead to early death. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition.

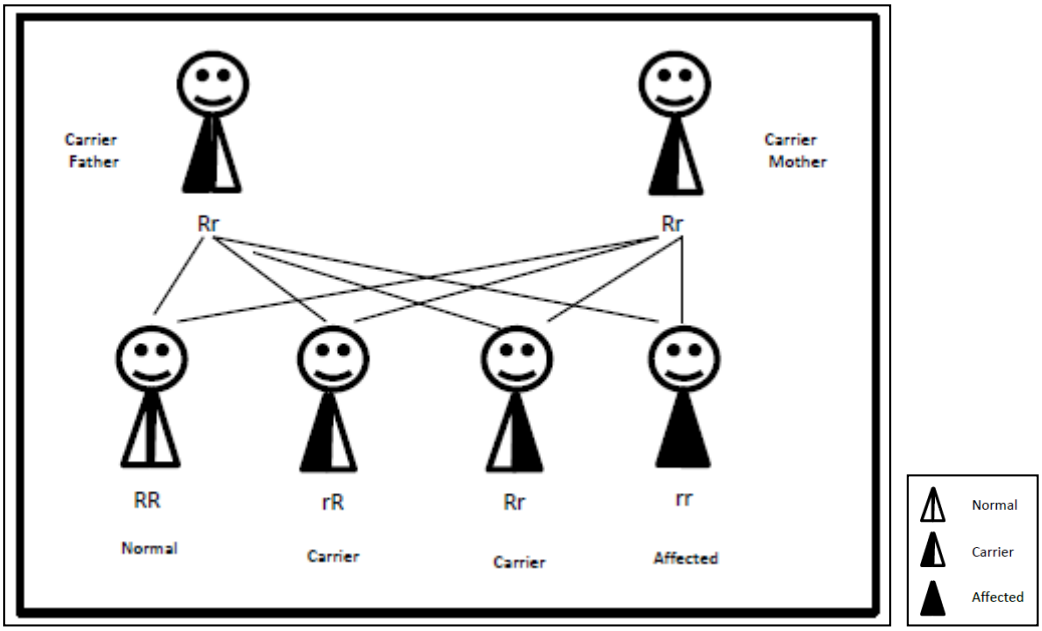
What causes Cystic Fibrosis?

It occurs when a particular cell protein called "cystic fibrosis transmembrane conductance regulator" (CFTR) is either missing or not working well. CFTR gene provides instructions for making a channel that transports chloride ions into and out of the cells. Mutations in CFTR cause disruptions in the flow of chloride ions and water across cell membranes. Cells that line passageways of lungs, pancreas, etc produce unusually sticky and thick mucus.

Cystic Fibrosis is an inherited condition. It affects both boys and girls equally. Everyone has a pair of genes that make the CFTR protein. In children with CF, neither of these genes works correctly. These children inherit one non-working gene for the condition from each parent.

Parents of children with CF rarely have the disorder. Instead, each parent has a single non-working gene for CF. They are called carriers. Carriers do not have CF because the other gene of this pair is working correctly.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have CF. There is a 50% chance for the child to be a carrier, just like the parents. And, there is a 25% chance for the child to have two working genes.



What are the signs and symptoms of Cystic Fibrosis?

Symptoms usually start in early childhood. In fact, most children with CF show effects before one year of age. There are some people who do not find out they have CF until adulthood.

Children born with this condition may have salty sweat (many parents notice a salty taste when kissing their child), poor weight gain and growth (even when a baby or child eats a lot), constant coughing, thick mucus and phlegm, many lung and sinus infections (pneumonias and bronchitis), greasy, smelly stools that are bulky and pale colored, intestinal problems (diarrhea or constipation, pain, gas) and polyps in the nose.

What is the treatment for Cystic Fibrosis?

The main goal of treatment is to keep the lungs clear of thick mucus and to provide with the correct amount of calories and nutrients to keep the patient healthy - vitamin supplements, a higher-calorie diet and extra fluid.

Other important Considerations

Have the child vaccinated according to the regular childhood schedule. Children with CF need all the usual childhood vaccinations. It is especially important to have a measles vaccine. In addition, the primary physician may suggest that the child have vaccinations against influenza and pneumonia on a yearly basis. Children with CF should also be protected against RSV, a respiratory illness that can be severe, and sometimes life-threatening, in children with chronic lung disease.

Keep the child away from all forms of smoke, especially cigarette smoke. It can add to lung damage.

Teach good hand washing habits to prevent infection.

If the child has a respiratory infection and is too sick to eat or follow regular health habits, call the attending physician right away. During some illnesses, the child may need to be seen in the hospital for treatment.

Encourage the child to get plenty of exercise. This will help maintain the child's lung function and improve overall health.

FATTY ACID OXIDATION DISORDERS [FAOD]

What are FAOD?

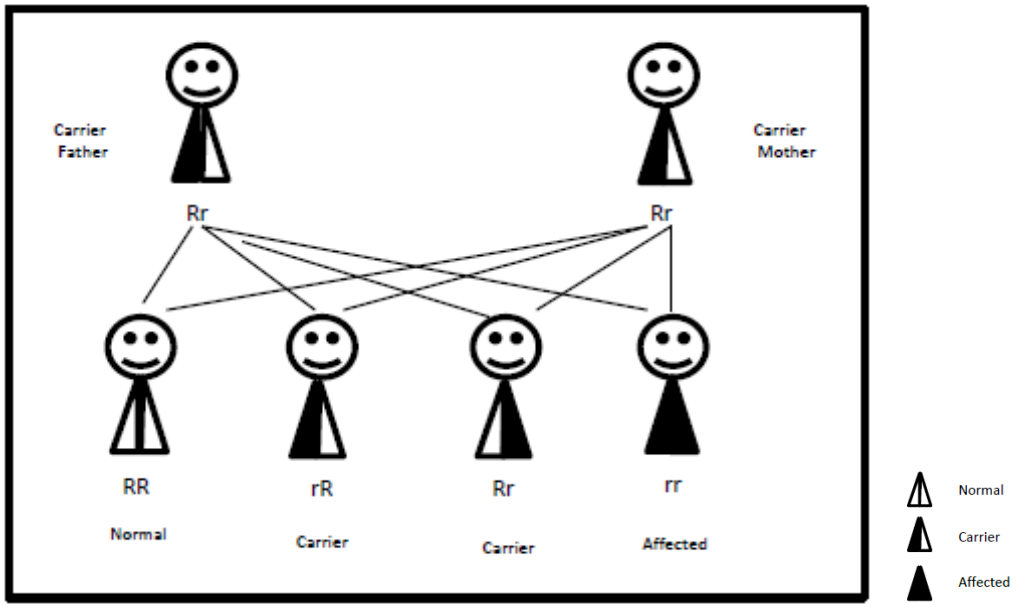
Fatty Acid Oxidation Disorders are a group of conditions due to a deficiency in an enzyme or chemical scissor that breaks down fats in the body. Children with this condition cannot properly breakdown fat in their body or from the food they eat. Children born with this condition appear normal at birth but if treatment is not given early, they may present with low blood sugar and lead to seizures, coma and death. These group of disorders can be detected through newborn screening. Early detection can prevent the complications of this condition such as low blood sugar, increased sleepiness, muscle weakness, heart problems among others.

What causes FAOD?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissor, children with this condition cannot effectively utilize fat from their body or from the food they eat.

FAOD are an inherited condition. The gene for the different enzymes is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with FAOD have one working and one non-working gene coding for a particular enzyme needed in the breakdown of fat. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have a FAOD. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of FAOD?

Children born with this condition appear normal at birth but if treatment is not given early, they can present with low blood sugar which can lead to seizures, coma and death. Having an enlarged heart or muscle weakness may be among the complications too.

What is the treatment of FAOD?

The main treatment of a FAOD is through control of the diet. If the child is well or does not have any illness, he/she should eat regular meals and avoid fasting more than 6 hours. One type of FAOD called VLCAD (or very long chain acyl-CoA dehydrogenase deficiency), will require a special milk formula called MCT (medium chain triglycerides).

What should I do when my baby is unwell or has an illness (like respiratory or gastrointestinal infection)?

Children with FAOD may have a "**metabolic crisis"** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drank well or during stressful events (such as surgery and severe infection). Your child may present with lethargy, seizures or convulsions, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

GALACTOSEMIA

What is Galactosemia?

Galactosemia for is one of the conditions that can be detected through newborn screening. Untreated children may develop cataracts, liver failure and developmental delay. Early detection can prevent the complications of this condition.

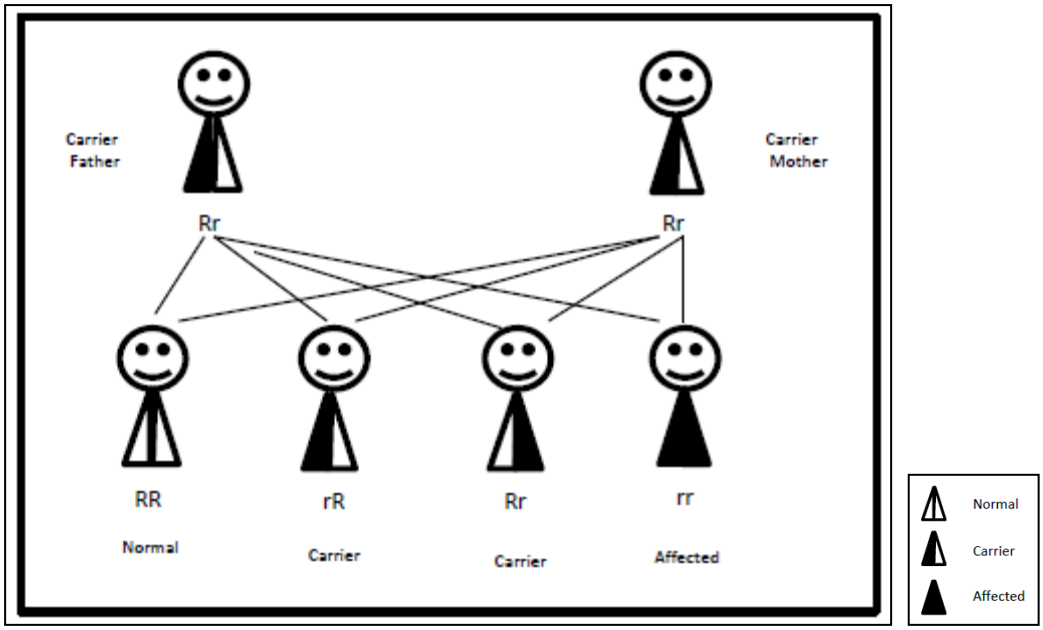
What Causes Galactosemia?

To efficiently use the food we eat, our body breaks these down to smaller units. Carbohydrates are broken down into sugars. A milk sugar called lactose is primarily affected in galactosemia. Due to a lack of an enzyme or chemical scissors, lactose cannot be broken down. This causes a sugar known as galactose to accumulate in the body.

The chemical process to break down galactose is complicated and involves several enzymes. Whether your child has classical or non-classical galactosemia will depend on which enzyme is deficient or missing. You may consult your doctor to know which type of galactosemia your child has.

Galactosemia is an inherited condition. The gene for the enzyme is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with Galactosemia have one working and one non-working gene coding for the enzyme. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have Galactosemia. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of Galactosemia?

Upon starting feeding with milk, the baby with galactosemia may have vomiting, irritability, yellow tinge of the skin (jaundice), severe infection (sepsis), enlarged liver and signs of liver failure such as bleeding. After a few weeks, patients may have cataracts. Without treatment, patients may die of liver failure or have cataracts and developmental delays.

What is the treatment of Galactosemia?

The main treatment of Galactosemia is through control of the diet. Because children with galactosemia cannot efficiently breakdown lactose, treatment is through the elimination of lactose from the diet. Babies are given soy-based milk formula. As their diet progresses, food which contain dairy and lactose are avoided altogether. It is important that the diet be followed to avoid any complications.

The control of galactosemic patients are evaluated with the help of blood spots, similar to the samples taken during their newborn screening. The levels of galactose metabolites are measured. As part of their monitoring, children with galactosemia should have their levels taken regularly and visit their physicians to assess their growth and development.

GLUCOSE-6-PHOSPHATE DEHYDROGENASE (G6PD) DEFICIENCY

What is G6PD deficiency?

Glucose-6-phosphate dehydrogenase deficiency, or G6PD deficiency for short, is the most common enzyme deficiency worldwide. This is an x-linked inherited disorder which means that from the time a baby is born, there is already something wrong with how his body makes and breaks important substances. According to statistics, about 400 million people have G6PD deficiency, and it is most common in Africa, Southeast Asia and the Middle East.

Babies with G6PD deficiency have very little or no enzyme called Glucose-6-Phosphate Dehydrogenase (G6PD). An enzyme is a kind of protein that speeds up chemical reactions in the body. The enzyme G6PD is especially important to red blood cells. If this enzyme is lacking or missing, red blood cells are easily destroyed. Another name for G6PD deficiency is favism because some people who have it, usually those living in the Mediterranean region, react very badly after ingestion of fava beans.

What causes G6PD deficiency?

In order to understand what causes G6PD deficiency, one must first learn a bit about genes and chromosomes. Genes are like the body's blueprints. They contain instructions on how specific parts of the body are made. For example, if the instructions in your hair genes say your hair is black, your hair will be black. Genes are packaged into threadlike structures called chromosomes. A chromosome is very much like a beaded bracelet. The beads are the different genes that give instructions for different part of the body; the entire bracelet is the chromosome. Genes usually come and act in pairs. One member of a specific pair comes from the father, and the other member comes from the mother. The members of a pair are located on paired chromosomes.

All normal human beings have 23 pairs of chromosomes. Each of the first 22 pairs contains the same number and kind of genes. The last and 23rd pair is the sex chromosomes. They are different from the first 22 pairs in that they do not have the same number and kind of genes. The sex chromosomes contain the genes that determine whether a baby will be a girl or a boy.

There are 2 kinds of sex chromosomes, X and Y. All baby girls have two X chromosomes. All baby boys have one X and one Y. The gene that gives instructions on how G6PD is made is found in the X chromosome only, thus G6PD deficiency is described as X-linked.

If a baby girl gets one defective G6PD gene from either of her parents, she will not have G6PD deficiency because she has another G6PD gene that can do the work (remember: a baby girl has two X chromosomes, thus two G6PD genes). But if she gets two deffective G6PD genes from both her parents, she will have G6PD deficiency. On the other hand, a baby boy whose G6PD gene is defective will surely get G6PD deficiency because the Y chromosome has no G6PD gene. A defective G6PD gene will give wrong instructions on how to make the enzyme G6PD. As a result, too little or none of it is made.

What are the harmful effects of G6PD deficiency?

G6PD has a very small but strategic role in protecting the body from substances that can cause damage to cells

or oxidative substances. Because of this important role, G6PD is normally found in all parts of the body. To be sure, most parts of the body also keep a "spare" enzyme, one that can do the work of G6PD in case it is lacking or missing entirely. Unfortunately, this is not the case with red blood cells. They do not have spare enzymes that can do the work of G6PD. If a baby does not have enough G6PD, his red blood cells lack protection from the harmful effects of oxidative substances.

A baby with G6PD deficiency appears and remains healthy until he is exposed to a large amount of oxidative substances. When this happens, his red blood cells are destroyed, a process known as hemolysis. Red blood cells carry oxygen to all parts of the body. When they undergo hemolysis, the baby will have hemolytic anemia. The signs and symptoms of hemolytic anemia are paleness, dizziness, headache, tea-colored urine, and abdominal or back pain or both. Hemolytic anemia, when very severe, can end in death.

Destroyed red blood cells are brought to the liver to be broken down to smaller pieces for disposal. One of the end products of this process is bilirubin, a yellowish substance that accumulates in different parts of the body when too much of it is produced. Quite often, bilirubin accumulates in the skin and causes it to appear yellowish. In the worst cases, bilirubin accumulates in the brain and causes mental retardation or death.

Where do oxidative substances come from?

Hemolysis of red blood cells will only occur IF and WHEN a G6PD deficient child is exposed to oxidative substances. Oxidative substances are found in certain drugs, foods, and beverages. The body also produces oxidative substances during severe infections or illnesses such as typhoid fever, pneumonia, or kidney failure. Most drugs with strong oxidative effects are of kinds:

1. antibiotics of the sulfa group
2. medicines for malaria
3. some medicines for fever

How is G6PD deficiency treated?

When a child has taken oxidative substances and suddenly shows the signs and symptoms of hemolytic anemia, he is said to have a hemolytic crisis. During such crisis, the goal of doctors and nurses is to prevent the harmful effects from getting worse. Blood transfusion, oxygen, and folic acid may be given. The ultimate treatment for G6PD deficiency is gene therapy (replacing a defective gene with a good one), but this is not yet available at the present time.

As parent, what should I do to prevent a hemolytic crisis?

1. Tell your child's pediatrician that your child has G6PD deficiency. This is very important so that he will not prescribe oxidative drugs in case your child gets ill. He would also be able to watch out for hemolytic crisis and would immediately know what to do just in case it happens.

2. Keep your list of oxidative substances in a handy place. Better yet, post it in a convenient spot on the kitchen wall. Always double-check food, beverage, and medicine labels against the list.

3. Memorize the signs and symptoms of hemolytic anemia: paleness, dizziness, headache, difficulty in breathing, rapid and strong heartbeats, tea-colored urine, and abdominal or back pain. Bring your child to his pediatrician as soon as these signs and symptoms appear.

4. Do not ignore infections. Persistent fever signals an infection. Bring the child at once to his pediatrician.

5. As your child gets older, honestly and gently tell him about his condition and teach him to be careful about what he eats.

My baby did not go through newborn screening. What should I do to make sure that he/she is not G6PD deficient?

You can bring your child to a pediatrician for a thorough medical checkup. If the age of your baby is already beyond the limit for newborn screening, he/she can still undergo confirmatory testing for G6PD deficiency. However, for the other diseases included in the newborn screening, it will depend on the assessment of your pediatrician if your baby will need a referral to an endocrinologist or geneticist for further evaluation.

My baby has G6PD deficiency and I feel devastated. Did I do something wrong when I was pregnant? Could I have done something while I was pregnant for my baby to avoid it?

You did nothing wrong. It is a genetic disorder, hence, there is nothing you can do as a mother to prevent it.

My firstborn was diagnosed with G6PD deficiency. I am pregnant again and worried – will my next baby also have it?

Once you have a child with G6PD deficiency, there is always a possibility of having a child with G6PD deficiency for every succeeding pregnancies. All your babies must undergo newborn screening.

Is it possible that the confirmatory test of my child is negative?

Yes. If negative, then you do not have to worry about the medicines and the food items. If positive, make sure that your attending physician knows that your child has G6PD deficiency.

I am a parent. Should I get tested for G6PD deficiency?

Yes, you may want to have yourself tested by requesting for G6PD assay using the confirmatory test in designated confirmatory centers. However, only males who are G6PD deficient and affected females (homozygous for G6PD) can be identified. A female with a carrier status (heterozygous for G6PD) cannot be detected by this kind of test.

I am trying to conceive. Should I be tested for G6PD deficiency?

It is your choice if you want to be tested but the probability of having a child with G6PD deficiency does not solely depend on your status. Your spouse's G6PD status can also affect the probability of having a child with G6PD. It will be more practical to just have your new baby undergo the routine newborn screening after birth.

Where is G6PD Deficiency Confirmatory Testing done?

At the moment, there are 15 Confirmatory Testing Centers in the country. You may want to contact them directly regarding their testing schedule. Please visit www.newbornscreening.ph for details.

Is formula milk safe for my son who has G6PD deficiency?

Although soya and soy lecithin are both included in the list, many products contain very small amount of soya including the common milk formulas. There has not been any report of hemolysis due to milk products containing soya. Soy lecithin, which is present in different brands, is an important component of milk but it does not make up the whole milk. **REMEMBER: Breastfeeding is still best for babies. Breastfeeding is healthier for both babies and mothers.**

I am breastfeeding my son. When I haven't received the newborn screening result I ate the foods that need to be avoided. Will they take effect on my son?

Theoretically, food and chemicals can be excreted through the milk. If your son did not develop hemolysis due to that event in the past, then probably they were not excreted at all through the milk or the amount was too small to cause significant reactions.

I have donated blood several times without knowing I am a G6PD carrier. Can I still donate blood?

Yes, you may still donate. There is no contraindication as a person with G6PD deficiency. There is also no policy written that you cannot donate.

Is it transmissible via blood transfusion?

No, it is not transmissible via blood transfusion.

My baby has G6PD deficiency; vitamins are on the restricted list. How can I keep him healthy?

Multivitamins are generally not contraindicated. Vitamin C or ascorbic acid can be given for as long as the dose is within the recommended dosage. Multivitamins are considered food supplements, but there is no substitute to nutritious, freshly prepared food and a well-balanced diet.

Is it okay to eat chocolates even they have soy content?

Although soya is included in the list, many products contain very small amount of soya. So generally patients with G6PD deficiency can eat chocolates except if it is a fava bean coated with chocolates. Please read the product label.

Is malunggay leaves safe to eat? Because I read in some website that malunggay isn't safe for people with G6PD deficiency?

For as long as it is not included in the official list of NIH as foods to avoid, then there is no basis to withhold malunggay from persons with g6pd.

I have a son who has G6PD deficiency and now he has a cold and cough, it is safe to use oregano for his medication?

Since oregano is not included in the list it can be given. However, you should exercise caution in giving commercially produced herbal preparations because they might contain unreported chemicals. Generally, cough and colds can be treated with lots of fluids. If proven to be bacterial in origin, they should be given antibiotics.

Is infant cereal safe for newborns with G6PD deficiency?

So far, there are no reported cases of hemolysis secondary to rice infant cereals (eg Cerelac). It contains a very small amount of soya in the first place.

Is insect repellant safe to use?

If you are not comfortable in using insect repellant, you can look for an organic type . If not available, you can just apply the commercial ones on your baby's clothes.

What toothpaste brand is safe for babies with G6PD?

The amount of menthol that is in ordinary toothpastes so far has not caused hemolysis in G6PD deficient patients.

What about blueberries? In other countries, they are not listed as contraindicated food.

If there is confusion about blueberries then it might be best to avoid it until extensive evidence that it is safe for patients with G6PD deficiency becomes available. Please note that patients with G6PD deficiency are not the same all over the world because of differences in mutations and therefore phenotypes, some of them will react consistently with some of the items in the list and some would not. As an organization, we are just giving the parents some guidance on what to avoid for the good of their children. It is still up to the parents to accept our recommendations.

What are the signs of hemolysis?

Signs of hemolysis include tea colored urine, yellowish discoloration of the skin, weakness and irritability. A child may look clinically pale but may actually have normal CBC results. So a CBC should be requested to confirm if the child has anemia.

-

My son exhibited symptoms of a viral infection (vomiting, lbm, lack of appetite). I saw on his diaper that the stain of his urine is somewhat tea colored. Should I be worried? What should I do?

Bring him to his pediatrician for checkup. Please do CBC APC, and urinalysis. Check for urine hemoglobin.

Have there been reported cases of hemolysis here in the Philippines because of G6PD deficiency? If so, what were the most common causes?

Yes, there have reports of hemolysis among patients with G6PD deficiency. There are also claims registered at PhilHealth. Some of the causes of hemolysis recorded in Philippine Children's Medical Center are infections (Flu, hepatitis) and exposure to moth balls. Fortunately, none of these patients died because of severe anemia. They were brought immediately to the hospital as soon as the initial signs of hemolysis became evident

What are the chances of getting a different result if we undergo another Confirmatory Test?

The family of a child confirmed with G6PD deficiency may always opt to have another confirmatory test if they are having doubts. If the result is below the borderline, the patient will most likely get a positive result in another test.

As a mom, can I use menthol-containing products?

The use of menthol and camphor containing products are not recommended if the goal is to reduce or eliminate the pain. These products only divert attention away from the painful area by producing cooling effects and sensation on the skin. It is always better to know the real cause/s of the pain/discomfort by consulting your doctor about it.

Is there a contraindication to any vaccination for patients with G6PD deficiency?

None.

IMPORTANT REMINDERS FOR PATIENTS WITH G6PD DEFICIENCY

1. Avoid ingestion of or exposure to the listed drugs and chemicals.
2. If you have coughs, cold or other bacterial or viral infections, make sure to inform your doctor that you have G6PD eficiency.
3. If you have ingested or were exposed to any medication and your urine became tea-colored, inform your doctor immediately.
4. If you have yellowish discoloration of skin, sclera or any part of your body, consult your doctor Immediately.

I. DRUGS TO BE AVOIDED	
Generic Name	**Common Brand Names**
A. . Antibacterial	
**Nalidixic acid*	
Nitrofuran 1. nitrofurantoin 2. furazolidone 3. nitrofurazone / nitrofural	Macrodantin Diafuran, Diapectolin, Furoxone Furacin
**P- aminosalicylic acid*	
B. Analgesic/ Antipyretic	
**Acetanilid*	
C. Antihelmentic	
**B-naphthol*	
**Niridazole*	
**Stibophan*	
D. Sulfonamides and Sulphones	
Dapsone	Lepravir
**Glucosulphone sodium*	
Glyburide/ Glibenclamide	Euglucon Gluban Lodulce Orabetic
**Mafenide acetate*	
**Salicylazosulphapyridine/ Sulfasalazine*	
Stibophen	(2-(2-Oxido-3,5-Disulphonatophenoxy)-1,3,2,Benzodioxastibole-4-6-Disulphonate)
Sulphacetamide/ Sulfacetamide	Cetapred Sensocet
**Sulphadimidine*	
**Sulphafurazone*	
Sulphamethazole/ Sulfamethazole	Bacidal Bactille Forte Bactrim Bacxal DLI Cotrimoxazole Forteprim Globaxol Pharex Cotrimoxazole Ritemed Cotrimoxazole Septrin Trim S
Sulphanilamide/ Sulfanilamide	
Sulphapyridine	
**Sulphoxone/ Sulfoxone*	
Sulfasalazine, Salazosulfapyridine	Salazopyrin
E. Antimalarials	
Chloroquine	Aralen Chlorofoz
**Pamaquine*	
Primaquine	
Pentaquine	
F. Miscellaneous	
Acetylphenylhydrazine	
Dimercaprol	
Futamide	
Isobutyl nitrate	
Mepacrine	
Phenazopyridine	Azomir
Probenecid	
Thiazolesulfone	
Urate oxidase/ Rasburicase	

II. CHEMICALS TO BE AVOIDED	
Methylene Blue	
Arsine	
Phenylhydrazine	
Toluidine blue	
Trinitrotoluene	
Aniline dyes	

III. FOOD AND DRINKS TO BE AVOIDED	
Fava beans	Dingdong nuts, Mr. Bean
Red wine	
Legumes	Abitsuelas, Garbanzos, Kadyos, Munggo
Blueberry	
Soya food	Taho, Tokwa, Soy Sauce
Tonic water	
Bitter melon / ampalaya	

IV. AT IBA PA	
Menthol	Alaxan Gel Ben-gay Efficascent Oil
	Listerine mouthwash Listerine Pocketpacks Megascent Oil Mentopas Medicated Plaster Omega Pain Killer
Camphor	
Naphthalene	Moth balls
Henna	
Herbs	Cattle gallstone bezoar Honeysuckle flower Chimonanathus flower 100% pearl powder Figwortflower Acalypha indica

V. DRUGS SAFE TO TAKE IN THERAPEUTIC DOSES	
Acetaminophen	Paracetamol, tylenol
Acetophenetidin/ phenacin	
Aspirin/ Acetylsalicylic acid	Alka-seltzer Aspilets Cor-80 Cortal
Ascorbic acid	
Chloramphenicol	Chlormycetin Chloro-S Chlorsig Klorfen Oliphenicol Optomycin Pediachlor Penachlor Speradex
Ciprofloxacin	Ciprobay CIpromax Cipromet Qinosyn-500 Quilox Xipro
Diphenhydramine	
Isoniazid	
Phenytoin	
Quinidine	
**Vitamin K analogues/ Phytomenadione	Hema-K Konakion MM Phil Pharmawealth/ Atlantic Phytomenadione

**Not Available in the Philippines*

***Should be water soluble*

GLUTARIC ACIDURIA TYPE I [GA1]

What is GA1?

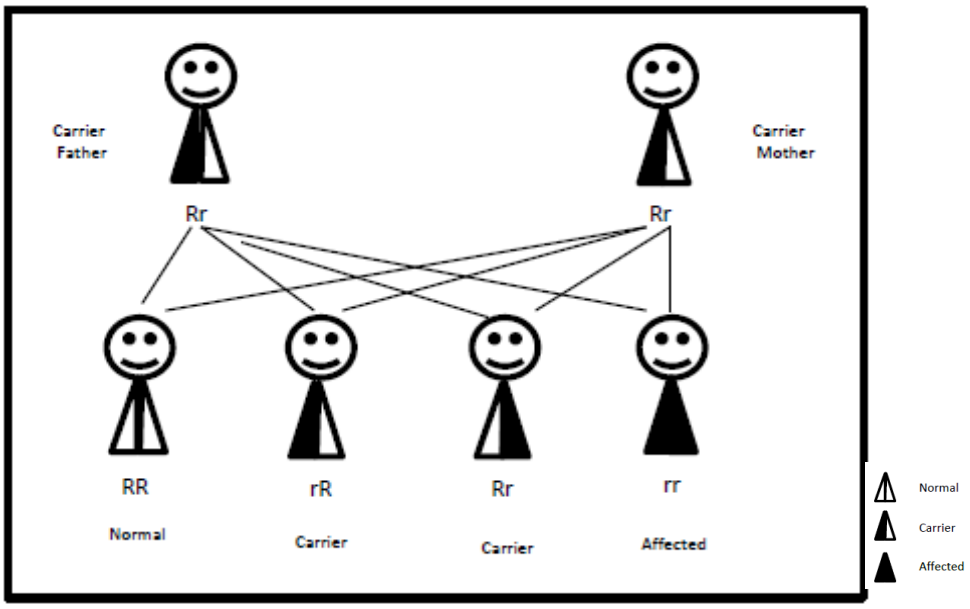
Glutaric Aciduria is due to a deficiency of an enzyme (or chemical scissors) called Glutaryl Co-A Dehydrogenase which is needed to break down the amino acids lysine and tryptophan. Amino acids are the building blocks of proteins. Children born with this condition may have an increased size of their head and may have convulsions, spasms of the muscles, involuntary movements of the arms or legs, coma and death. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition.

What causes GA1?

To efficiently use the food we eat, our body breaks down the amino acids to smaller units. Due to a lack of an enzyme or chemical scissors, children with this condition cannot effectively break down the amino acids lysine and tryptophan. The accumulation of glutaric acid, a by-product of these amino acids, causes the signs and symptoms of this condition.

The gene for the glutaryl-CoA dehydrogenase enzyme is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with Glutaric Aciduria Type 1 have one working and one non-working gene coding for this enzyme. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have glutaric aciduria type 1. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of GA1?

Children born with this condition may have an increased size of their head and may have convulsions, spasms of the muscles, involuntary movements of the arms or legs, coma and death.

What is the treatment of GA1?

The main treatment of GA1 is through control of the diet. The child's diet should be low in tryptophan and lysine. Tryptophan and lysine are contained in protein rich foods. Children with glutaric aciduria are given a special milk formula without tryptophan or lysine. They are also given carnitine, a medication that helps the cells in the body to make energy.

What should I do when my baby is unwell?

Children with GA1 may have a **metabolic crisis** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drank well or during stressful events (such as surgery, severe infection and vaccination). Your child may present with lethargy, seizures or convulsions, increased in involuntary movements, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

GLUTARIC ACIDURIA TYPE II [GA2]

What is GA2?

Glutaric Aciduria Type II is due to a deficiency in the electron transfer flavoprotein and/or the ETF-ubiquinone oxidoreductase enzyme or chemical scissor that breaks down fat and proteins in the body. The deficiency of one or both of the enzymes may cause Glutaric Aciduria type II. Children with this condition cannot properly breakdown fat and some amino acids (or the building blocks of protein) in their body or from the food they eat.

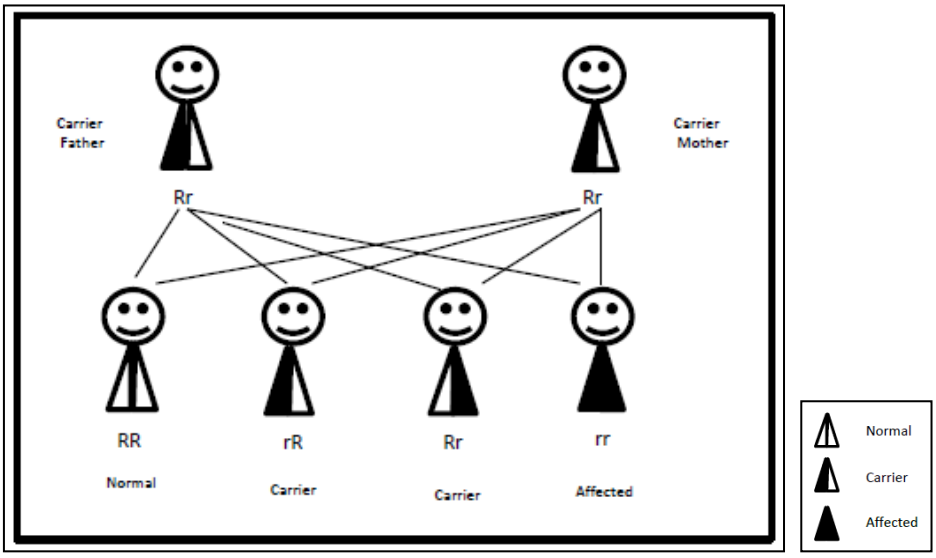
Some children born with this condition may not display any signs or symptoms until later in life but some may present at infancy with nausea, vomiting, weakness and low blood sugar which may lead to seizures, coma and death. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition such as low blood sugar, increased sleepiness, and muscle weakness among others.

What Causes GA2?

To efficiently use the food we eat, our body breaks fats and proteins down to smaller units. Due to a lack of an enzyme or chemical scissor, children with this condition cannot effectively utilize fat or some amino acids (or the building blocks of protein) from their body or from the food they eat.

Glutaric Aciduria Type II is an inherited condition. The gene for the enzyme is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with Glutaric Aciduria Type II have one working and one non-working gene coding for a particular enzyme needed in the breakdown of fat and some proteins. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have Glutaric Aciduria Type II. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of GA2?

Children born with this condition appear normal at birth but if treatment is not given early they can present with low blood sugar which can lead to seizures, coma and death. Muscle weakness may be among the complications too.

What is the treatment of GA2?

The main treatment of GA2 is through control of the diet. Children with this condition should aim for a diet high in carbohydrates and low in protein and fat. If the child is well or does not have any illness, he/she should eat regular meals and avoid fasting more than 6 hours.

Riboflavin, a vitamin, has been found to be helpful. Carnitine, a supplemental medication essential for muscle energy production may also be of benefit for some patients.

What should I do when my baby is unwell or has an illness (like respiratory or gastrointestinal infection)?

Children with GA2 may have a "**metabolic crisis"** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drank well or during stressful events (such as surgery and severe infection). Your child may present with lethargy, seizures or convulsions, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

HOMOCYSTINURIA

What is Homocystinuria?

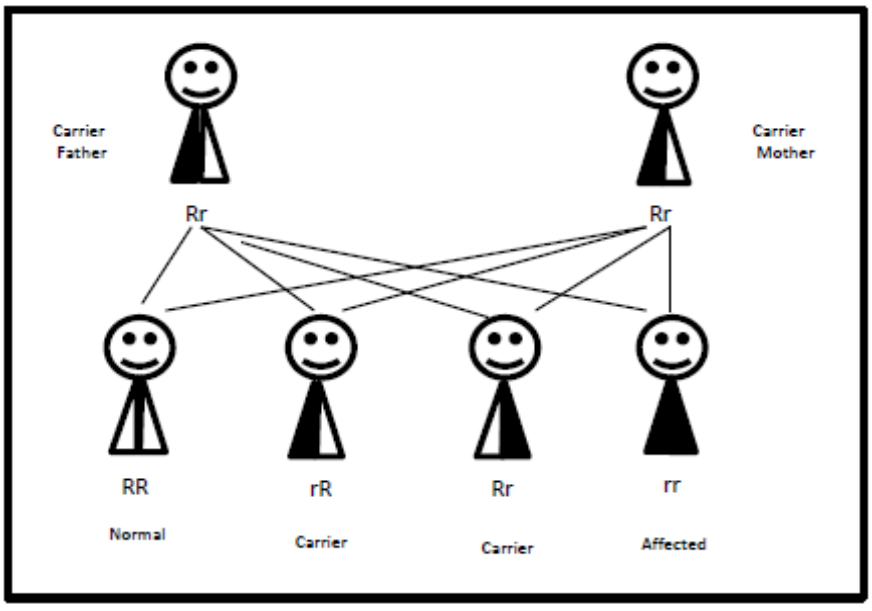
Homocystinuria is a condition where there is accumulation of methionine (an amino acid which is a building block of protein). Children born with this condition appear normal at birth but during childhood untreated children may present with visual problems, mental retardation, scoliosis and a higher risk of blood clotting abnormalities. It is one of the conditions that can be detected through newborn screening. Early detection can prevent the complications of homocystinuria.

What causes Homocystinuria?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors (in this case, the cystathionine B synthase enzyme), methionine (an amino acid) cannot be broken down. The accumulation of this amino acid causes the signs and symptoms of homocystinuria.

Homocystinuria is an inherited condition. The CBS gene is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with HCY have one working and one non-working gene coding for CBS. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have homocystinuria. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of Homocystinuria?

Children with homocystinuria appear normal at birth but during childhood untreated children may present with visual problems, mental retardation, scoliosis and a higher risk of blood clotting abnormalities.

What is the treatment of Homocystinuria?

The main treatment of homocystinuria is through control of the diet and intake of some vitamins. If the child is well or does not have any illness, he/she should continue taking the special milk formula and be on low protein diet as ordered by your pediatrician or metabolic specialist. It is important that the diet be followed to avoid any complications. In addition, vitamin B6, folic acid and a medication called betaine may help in breaking down excess methionine.

What are the precautions that I should take?

Children with homocystinuria may have a **metabolic crisis** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not drank well or during stressful events such as surgery. Your child may present with blood clotting abnormalities such as stroke. If your child is unwell or will undergo surgery, please bring him/her to the hospital for management and alert your pediatrician or metabolic physician.

MAPLE SYRUP URINE DISEASE [MSUD]

What is MSUD?

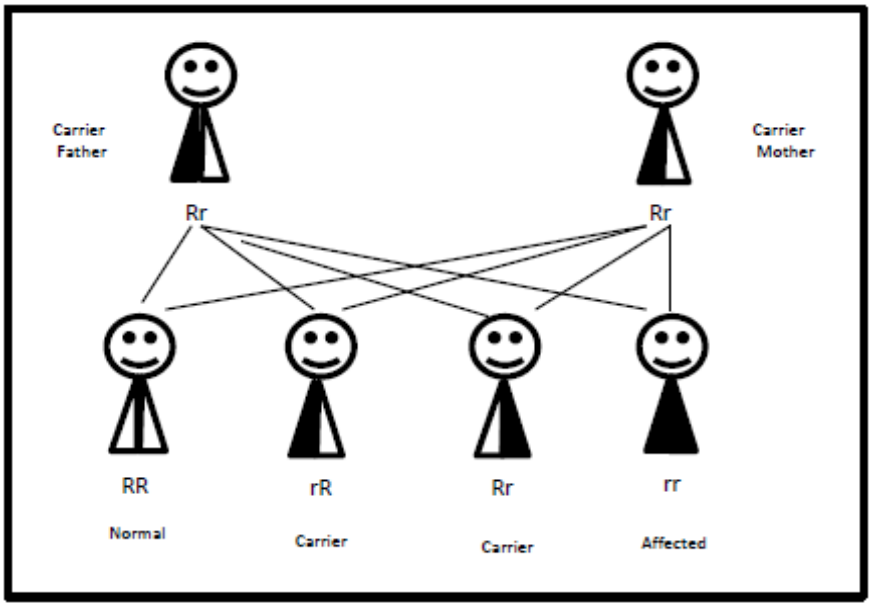
MSUD is short for maple syrup urine disease. Untreated children with this condition have a "sweet smelling" (similar to burnt sugar) urine from where its name is derived. In addition, untreated children may have complications such as convulsions, mental retardation and death. It is one of the conditions that can be detected through newborn screening. Early detection can prevent brain damage.

What Causes MSUD?

To efficiently use the protein we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors (in this case, the BCKAD enzyme), some amino acids which are the building blocks of protein cannot be broken down. The accumulation of these amino acids causes the signs and symptoms of MSUD.

MSUD is an inherited condition. The BCKAD gene is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the BCKAD gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with MSUD have one working and one non-working gene coding for BCKAD. They do not manifest the disease but can pass them on to their children. They are known as carriers.



If the child inherits the non-working gene from both parents, he or she will have MSUD. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of MSUD?

Children with MSUD may present with lethargy (or sleepiness), vomiting, poor suck, seizures and sweet smelling urine. Without treatment, patients may have swelling of the brain and coma. Brain damage can occur and mental retardation may ensue if patients are not treated well.

What is the treatment of MSUD?

The main treatment of MSUD is through control of the diet. If the child is well or does not have any illness, he/she should continue taking the special milk formula and be on low protein diet as ordered by your pediatrician or metabolic specialist. It is important that the diet be followed to avoid any complications.

What should I do when my baby is unwell?

Children with MSUD may have a **metabolic crisis** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drank well or during stressful events (such as surgery and severe infection). Your child may present with lethargy, seizures or convulsions, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

METHIONINE ADENOSINE TRANSFERASE DEFICIENCY

What is methionine adenosine transferase deficiency?

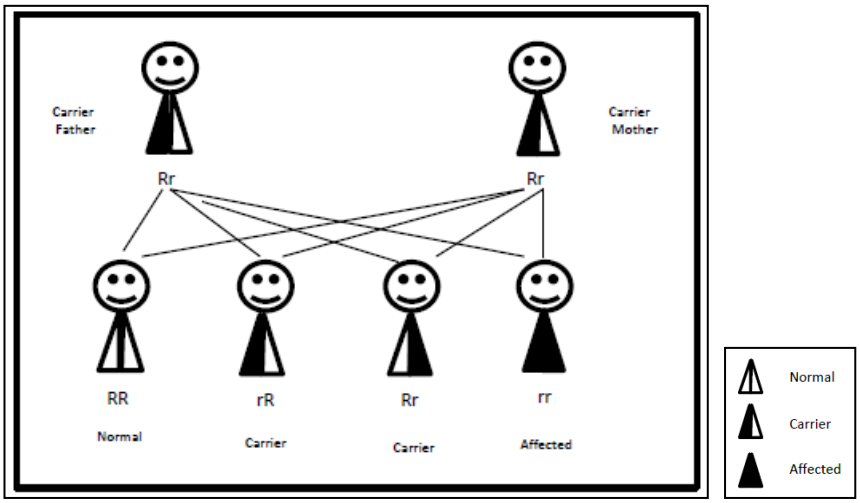
Methionine Adenosine Transferase (MAT) Deficiency is due to a deficiency in an enzyme or chemical scissor that breaks down the amino acid (or building block of protein) called methionine. Children with this condition cannot properly breakdown methionine in their body or from the food they eat. Children born with this condition appear normal at birth but if treatment is not given early, they may present with tremors, movement disorders, seizures and a "boiled cabbage" odor in the urine or sweat. This condition can be detected through newborn screening. Early detection can prevent the complications of this condition such as mental retardation and seizures.

What causes MAT Deficiency?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissor, children with this condition cannot effectively breakdown the amino acid methionine from their body or from the food they eat.

MAT Deficiency is an inherited condition. The gene for the different enzymes is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with MAT Deficiency have one working and one non-working gene coding for a particular enzyme needed in the breakdown of fat. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have a MAT Deficiency. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of MAT Deficiency?

Children born with this condition appear normal at birth but if treatment is not given early they can present with tremors, movement disorders and seizures. Mental retardation and death are among the complications.

What is the treatment of MAT Deficiency?

The main treatment of a MAT is through a low protein diet, specifically low methionine diet. S-adenosylmethionine administration has been found to be effective in neurologic development.

MULTIPLE CARBOXYLASE DEFICIENCY

What is Multiple Carboxylase Deficiency?

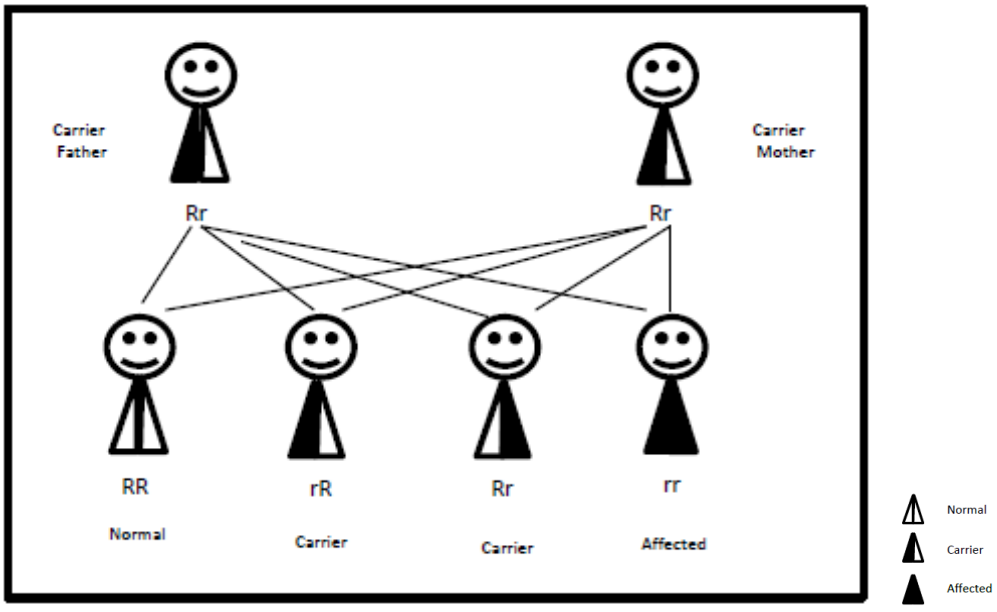
Multiple Carboxylase Deficiency is due to a deficiency of an enzyme (or chemical scissors) called holocarboxylase synthetase. Holocarboxylase synthetase adds biotin (a vitamin) to carboxylases which are other enzymes needed to change the food we eat into energy. Untreated children born with this condition may present with drowsiness, poor appetite, poor weight gain, vomiting, alopecia and a skin rash. They may have mental retardation and brain damage as a complication. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition.

What causes Multiple Carboxylase Deficiency?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors, children with this condition cannot effectively breakdown certain carbohydrates and fats which in turn are needed to breakdown protein.

The gene for the holocarboxylase synthetase enzyme is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with multiple carboxylase deficiency have one working and one non-working gene coding for this enzyme. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have multiple carboxylase deficiency. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

ORGANIC ACIDURIAS

What are Organic Acidurias?

Organic Acidurias are a group of conditions due to a deficiency in an enzyme or chemical scissor that breaks down proteins in the body. If not treated early, children with this condition may present with vomiting, irritability, drowsiness, rapid breathing and coma.

The name of the condition is derived from the substance that builds-up due to the deficiency of the said enzyme. An organic aciduria can be one of the following:

Propionic aciduria – due to a deficiency of the enzyme propionyl-CoA carboxylase
Methylmalonic aciduria– due to a defiency of the enzyme methymalonyl-CoA mutase
Isovaleric aciduria – due to a deficiency of the enzyme isovaleryl-CoA dehydrogenase

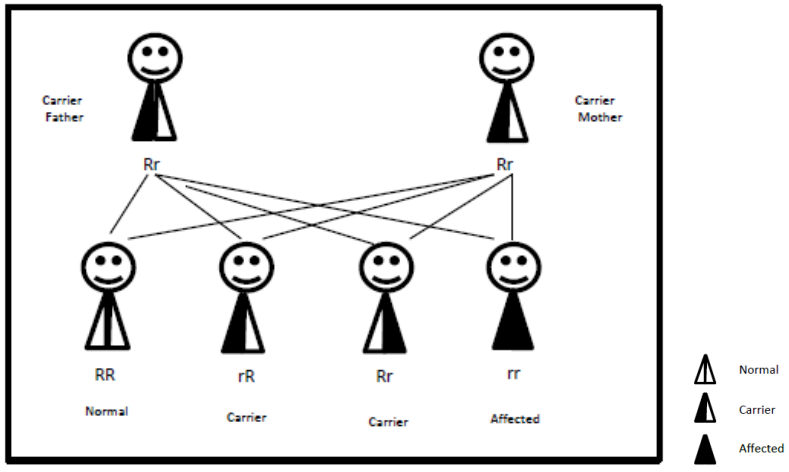
These group of disorders can be detected through newborn screening. Early detection can prevent the complications of these conditions.

What causes Organic Acidurias?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors, children with this condition cannot effectively breakdown protein which causes the accumulation of toxic substances in the body.

The organic acidurias are an inherited condition. The gene for the different enzymes is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with organic acidurias have one working and one non-working gene coding for a particular enzyme needed in the breakdown of protein. They do not manifest the disease but can pass them on to their children. They are known as carriers.



If the child inherits the non-working gene from both parents, he or she will have an organic acidemia. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of Organic Acidurias?

Untreated children with this condition may present with vomiting, irritability, drowsiness, rapid breathing and coma. They may become seriously ill and it may cause death.

What is the treatment of Organic Acidurias?

The main treatment of organic acidurias is through control of the diet. Children are placed on a low protein diet. They are also given carnitine, which is a medication that aids in the excretion of the toxic substances. Children with isovaleric aciduria are also given glycine which works similarly to carnitine. In some cases of methylmalonic aciduria, giving vitamin B12 is beneficial to the child.

What should I do when my baby is unwell or has an illness (like respiratory or gastrointestinal infection)?

Children with organic acidurias may have a "**metabolic crisis"** which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drank well or during stressful events (such as surgery and severe infection). Your child may present with lethargy, seizures or convulsions, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

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PHENYLKETONURIA

What is Phenylketonuria?

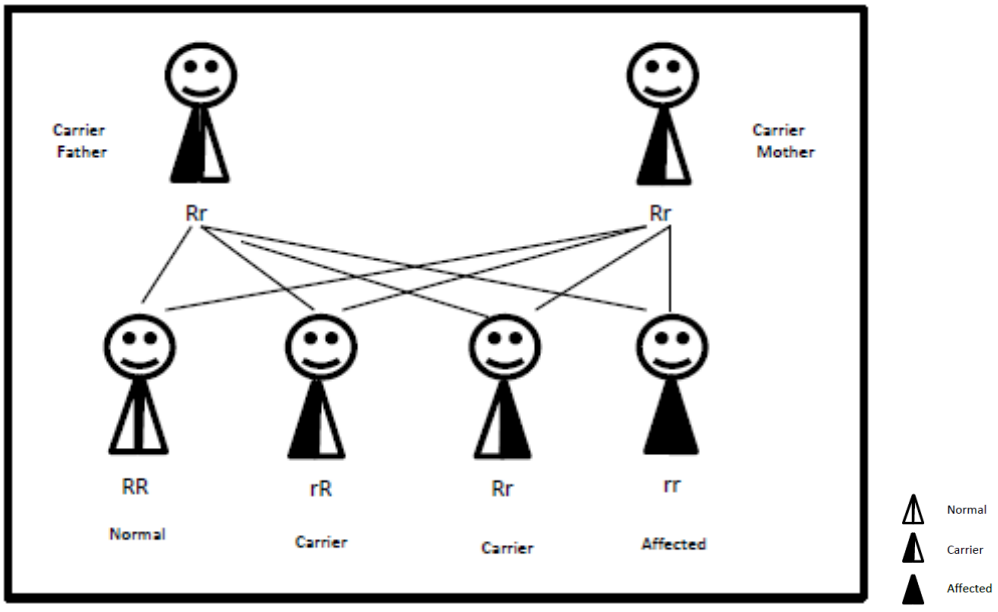
Phenylketonuria or PKU for is one of the conditions that can be detected through newborn screening. Untreated children may have mental retardation. Early detection can prevent brain damage.

What causes Phenylketonuria?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors (in this case, the phenylalanine hydroxylase (PAH) enzyme), phenylalanine, an amino acid which is one of the building blocks of protein cannot be broken down. The accumulation of phenylalanine causes the signs and symptoms of PKU.

PKU is an inherited condition. The PAH gene is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the PAH gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with PKU have one working and one non-working gene coding for PAH. They do not manifest the disease but can pass them on to their children. They are known as carriers.



If the child inherits the non-working gene from both parents, he or she will have PKU. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of PKU?

Children with PKU may seem normal at birth. They begin to manifest at about 6 months of age with delayed development (late in learning to sit, stand etc). Untreated children may also have decreased attention span, mental retardation, irritability, convulsions, hyperactivity and behavioral problems.

What is the treatment of PKU?

The main treatment of PKU is through control of the diet. If the child is well or does not have any illness, he/she should continue taking the special milk formula and be on low protein diet as ordered by your pediatrician or metabolic specialist. It is important that the diet be followed to avoid any complications.

Take note that sugar substitutes or artificial sweeteners (such as Equal or Splenda) may contain aspartame. Aspartame contains a high amount of phenylalanine. It is important that the child avoid foods or processed foods containing aspartame.

What should I do when my baby is sick?

When children with PKU are sick, phenylalanine may accumulate in the blood. Your child may present with lethargy, seizures or convulsions, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

THALASSEMIA

What is THALASSEMIA Trait/Minor?

These are mild disorders of the red cells which may result in a slight anemia. In most cases, children are symptom free and will have normal growth and development.

Does this mean that my child will lead a normal life?

Yes. It is important to remember that your baby will only have the trait but will not exhibit the disease as well as the signs of the disease. This trait may be transmitted by the infant to their would be children in the future so it is extremely important to have their future partners screened for the trait as their union may result in a baby with severe case of thalassemia or even fetal death. Severe cases of thalassemia will require frequent transfusion for their entire life.

Are there any precautions that I need to do or medications I have to avoid giving my baby?

You should refrain from giving iron supplements. Vitamin C, Folic acid, Vitamin E and D are recommended which are available in most multivitamin preparations.

Which doctor or specialist should I bring my baby for regular check ups?

Since the trait is basically silent, your pediatrician may take care of their health needs. Occasionally you may need to visit a paediatric haematologist depending on your paediatrician's clinical assessment.

What immunizations should they receive?

They need to receive the usual recommendations of the Philippine Pediatric Society. It is recommended though that they also be given Pneumococcal, influenza and HIB vaccines.

TYROSINEMIA TYPE I

What is Tyrosinemia Type I?

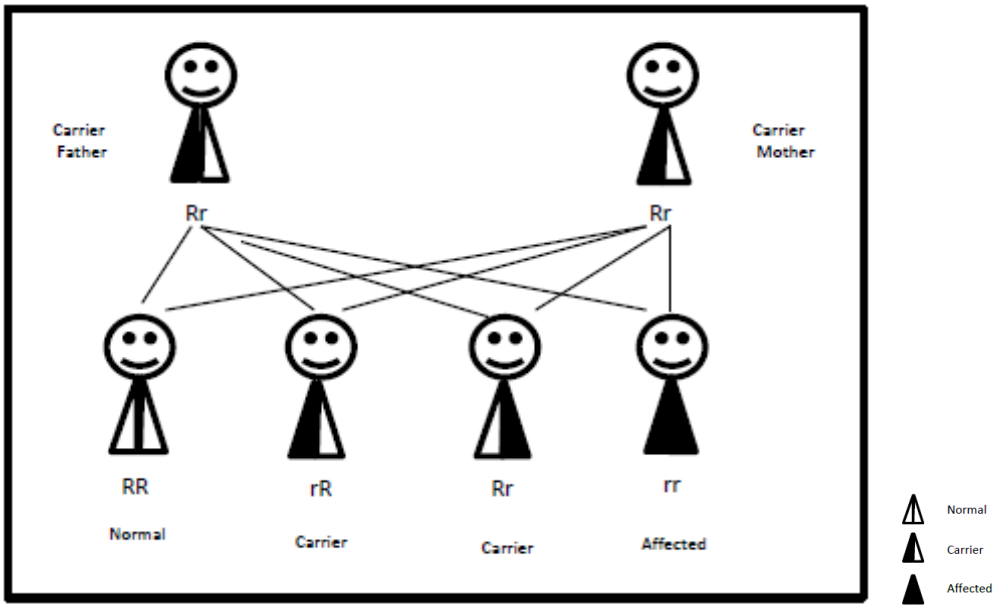
Tyrosinemia type I is a condition where there is accumulation of tyrosine (an amino acid which is a building block of protein) in the body. Due to this accumulation, another by product of tyrosine, succinylacetone and succinylacetoacetate also have increased levels in the blood. It is the rise of these chemicals that cause the signs and symptoms. Untreated children with this condition may have jaundice (yellowing of the skin), diarrhea and bloody stools, poor weight gain, irritability, drowsiness, enlarged liver and bleeding problems. Early detection can prevent liver damage.

What Causes Tyrosinemia Type I?

To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors (in this case, the fumarylacetoacetate hydrolase (FAH) enzyme), tyrosine (an amino acid) cannot be broken down. The accumulation of tyrosine, in turn, causes an increase in its by-products succinylacetone and succinylacetoacetate. It is the accumulation of these substances that causes the signs and symptoms of tyrosinemia.

Tryosinemia is an inherited condition. The FAH gene is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the FAH gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with Tryosinemia have one working and one non-working gene coding for FAH. They do not manifest the disease but can pass them on to their children. They are known as carriers.



If the child inherits the non-working gene from both parents, he or she will have Tyrosinemia Type I. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of Tyrosinemia Type I?

Children with Tyrosinemia Type 1 may present with jaundice (yellowing of the skin), diarrhea and bloody stools, poor weight gain, irritability, drowsiness, enlarged liver and bleeding problems. Without treatment, the liver can be damaged.

What is the treatment of Tyrosinemia Type I?

The main treatment of Tyrosinemia Type 1 is through control of the diet. If the child is well or does not have any illness, he/she should continue taking the special milk formula and be on low protein diet as ordered by your pediatrician or metabolic specialist. It is important that the diet be followed to avoid any complications. Patients are also given nitisinone (NTBC), which helps to control tyrosine levels.

What should I do when my baby is unwell?

When a child is sick, has not eaten/drank well during stressful events (such as surgery and severe infection) or has consumed a high amount of protein, there can be a build-up of toxic substances in the blood. Your child may present with yellowing of the skin, drowsiness, lethargy and bleeding. If not treated properly and immediately, it might lead to serious liver damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

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UREA CYCLE DEFECTS

What is the Urea Cycle?

When we eat food specifically proteins, our body breaks them down to smaller substances to be used efficiently for our body's function. Our body makes use of enzymes or chemical scissors to cut up proteins into building blocks called amino acids. We usually take in more protein than our body needs. The excess protein is then broken down into ammonia and organic acids. A high amount of ammonia in the body is dangerous and should be properly excreted. The urea cycle is the pathway for the ammonia to be processed and excreted by the body.

What is a Urea Cycle Defect?

A urea cycle defect occurs if there is a lack or absence of any enzyme within the cycle. While the presentation may be variable, it is one of the conditions that can be detected through newborn screening. Untreated children may have drowsiness, fast breathing, vomiting, irritability and they may die.

What is Citrullinemia?

Citrullinemia is a condition that results from a lack of the enzyme argininosuccinate synthetase. Argininosuccinate synthetase is one of the enzymes which are part of the urea cycle. The condition is called citrullinemia because citrulline (an amino acid) accumulates along with ammonia.

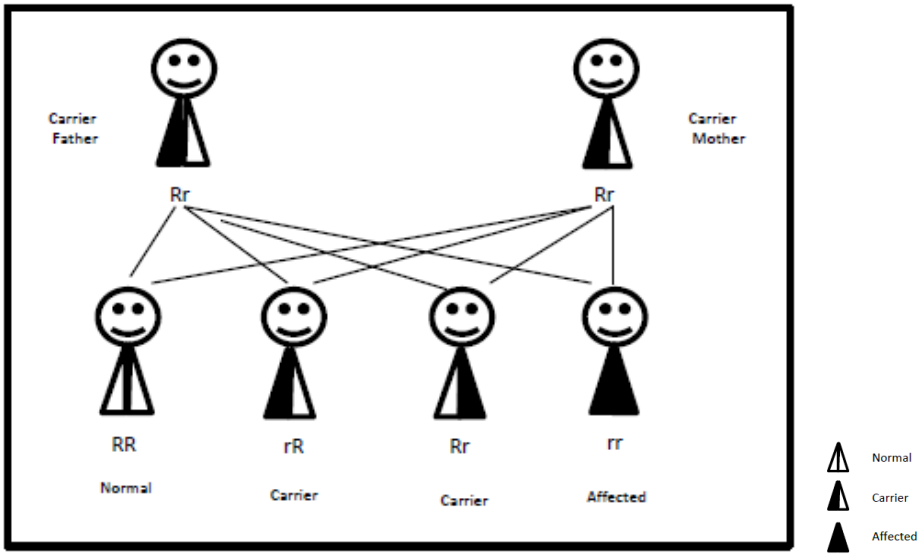
What is Arginosuccicinic Aciduria?

Argininosuccinate Aciduria (or ASA) is a condition that results from a lack of the enzyme argininosuccinate lyase. Argininosuccinate lyase is one of the enzymes which are part of the urea cycle. The condition is called argininosuccinate aciduria because arginosuccinate (an intermediate product of the cycle) accumulates along with ammonia.

What Causes Urea Cycle Defects (Citrullinemia and Argininosuccinic Aciduria)?

Citrullinemia and Argininosuccinic aciduria are inherited conditions. The genes coding for the enzymes are contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with this condition have one working and one non-working gene coding for the enzyme. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.



If the child inherits the non-working gene from both parents, he or she will have the condition. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of UCD?

Children with UCD may may have drowsiness, fast breathing, vomiting, convulsions, irritability and they may die.

What is the treatment of UCD?

The main aim of treatment of urea cycle defect is to keep the ammonia levels in the blood low or normal. To achieve this, patients with UCD are advised a low protein diet and to take a special milk formula. Sodium benzoate, a medication that helps in getting rid of excess ammonia, is also prescribed. Because children with UCD are found to have low levels of arginine (an essential amino acid), they are also given arginine supplementation.

What should I do when my baby is sick? (will undergo surgery or has an infection i.e. respiratory or gastrointestinal)

When children with UCD are sick, ammonia may accumulate in the blood. Your child may present with drowsiness, vomiting, seizures or convulsions and irritability. If not treated properly and immediately, it might lead to serious brain damage. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.