

UNDERSTANDING MAPLE SYRUP URINE DISEASE

What is maple syrup urine disease?

Maple syrup urine disorder is genetic disorder that affects how protein is broken down in the body. It is a **metabolic** disorder.

Amino acids are the building blocks of protein. Normally, the protein we eat is broken down or "metabolized" in our bodies into amino acids and used for growth and tissue repair.

People who are born with maple syrup urine disease are unable to properly break down some amino acids in the food they eat. Specifically, they are unable to breakdown the branch chain amino acids: leucine, isoleucine and valine. This is because they are missing an **enzyme** in their cells. Some people with maple syrup urine disease have a bit of this enzyme and some have none at all.



Enzymes can best be described as keys that unlock doors in the body. Think of the food in our system as a person with a big delivery to make. As long as the person has all the keys, the load can be delivered and everyone gets their packages. When enzymes or keys are missing, the packages cannot be delivered and the delivery person is left with a big load, and no one gets the packages they need to do their job.

Food eaten is like the delivery. In maple syrup urine disease, when the enzyme is missing, protein cannot be fully broken down for use by the body. Because these amino acids do not get broken down completely, high levels accumulate in the blood, urine and sweat. The byproduct of isoleucine has a characteristic sweet smell, which gives the disorder its name. The leucine, isoleucine and valine are like the delivery person's packages. They cannot be delivered to the right place and the pile just keeps growing.

When certain acids build up in the blood, they are like poison in the body. The person may not have an appetite, feel confused, not think clearly, sleep

more than usual, or vomit. If the extra amino acids are not removed quickly, the person may have seizures or may go into a coma.

In untreated maple syrup urine disease, the balance is upset between having enough protein (to meet body needs) and having too much protein (which causes toxic levels of acid in the blood and body tissues). This imbalance is called **metabolic acidosis** and can cause:

- stunted growth
- problems with the pancreas, or 'pancreatitis'
- bone marrow suppression causing low blood counts, easy bleeding and vulnerability to germs such as colds and flu
- osteoporosis, or thinning of the bones
- swelling around the brain
- brain damage
- seizures
- coma
- eye complications

Some people with maple syrup urine disease can have all of these complications and be severely affected whereas others may have only a few complications. It all depends on how much of the enzyme is working in their bodies and if their disorder is well-controlled.

How is maple syrup urine disease treated?

The treatment for maple syrup urine disease is aimed at preventing toxic high levels of leucine (an amino acid) in the body. Therapy needs to begin before complications occur because, once they develop, they are irreversible. Examples of this are seizures and mental retardation from swelling around the brain. Treatment must be started as soon as the disorder is diagnosed.

Treatment for Maple Syrup Urine disease includes:

DIET



People who have maple syrup urine disease need to strictly follow a special diet to stay well.

The first part of treatment is **reducing protein in the diet**. If less protein is taken in, less acid will accumulate.



Protein in the diet is lowered by avoiding protein-rich foods like meat and milk. Examples of low protein foods are fruits, vegetables and starches. These foods provide calories without loading the body with protein. Special low protein foods are also a key part of this diet.

Calories are a very important part of the diet. The body can use them for fuel without breaking down its own reserves. If the body does not have adequate fuel in the form of calories, it will actually begin to *break down muscle* to provide energy. Muscle is protein and will increase acid levels in the same way as eating protein will.

The metabolic dietitian works with people with maple syrup urine disease to ensure that adequate calories and nutrients are being provided while following this special low protein diet. The amount of food and drink the person can safely consume is calculated by the dietitian and often, all foods and fluids must be carefully measured to control the amount of protein consumed. The dietitian often requests that a record is kept of everything the patient eats and drinks so that adjustments can be made to the diet as needed.

Examples of foods that must be eliminated from a low-protein diet are:

- milk and dairy products
- meat, fish, chicken

- nuts (including peanut butter)
- beans
- eggs

Eating high protein foods increases acid levels. This puts the person at risk of losing consciousness, having seizures and for developing complications such as pancreatitis.

It is not unusual for someone on a low-protein diet to have 2 kinds of vegetables and a baked potato for dinner. However, if these foods were all that the person ate, their diet would be lacking in protein, vitamins and minerals. That is where the special **medical formula** comes in. It provides all the protein and nutrients normally obtained from high-protein food, but does not contain leucine, isoleucine and valine. Without this formula, the person with MSUD would be at high risk for many nutritional deficiencies. In order to prevent complications, the person must adhere to this diet for life.

The balancing act

The challenge in treating maple syrup urine disease is to provide enough protein to meet body requirements without overloading the person with protein waste (acid). The goal is to provide enough leucine, valine, and isoleucine to allow for growth but to prevent excessive intakes, which can make a person ill. Visits to the metabolic clinic are needed to consult with the team and make adjustments to diet and medication. Regular blood work is necessary to monitor changes in blood levels and other nutritional indicators.

The delicate balance can be upset by strenuous exercise and illness. Both can cause the acid levels to rise. Extra calories are needed at these times to provide the stressed body with fuel.

How do people get maple syrup urine disease?

Maple syrup urine disease is an inborn error of metabolism. It is genetic. This means that the person has the disorder from the time they are conceived. At conception, the baby receives two sets of genetic material, one from the mother, and one from the father. This genetic material, called **DNA**, acts as a recipe for the baby's development.

DNA includes information about the baby's eye and hair colour, sex and even whether the baby will be right or left-handed. DNA comes in units called **genes**. Each pair of genes gives directions to a certain part of the body.

In maple syrup urine disease, there is a problem with the genes that give information on how the body breaks down protein. The disorder is caused by getting two copies of the faulty gene that gives the wrong instructions. One copy comes from the mother and one comes from the father. If the child inherits only one copy of the gene, they are a carrier for maple syrup urine disease but are not affected. There is a 1 in 4, or 25% chance that two carriers of the gene will have a baby with maple syrup urine disease. Boys and girls are equally affected.

With each pregnancy, there is the **same** 25% chance of having a child with maple syrup urine disease. In some families, there may be only one child with maple syrup urine disease, while in other families, multiple children may be affected.



How can family and friends help?

1. Learn to recognize the signs of trouble. If you know the person well, you will be the first to notice changes in behavior that suggest the acid levels are too high. For example, the person may become irritable, less active and sleep more than usual. Encourage the person to call the clinic and have bloodwork drawn.

Very high levels of acid in the blood may cause problems like:

- confusion
- vomiting
- increased sweating
- laboured breathing
- loss of consciousness
- seizures

If your friend or family member show any of the signs or symptoms of high acid levels or **metabolic acidosis**, take them to the emergency room immediately. The person with the maple syrup urine disease has a yellow wallet card with emergency room instructions and phone numbers.

2. It is very hard to constantly be faced with food temptations when you are trying to stick to a special diet. Understanding the challenges of living with maple syrup urine disease and offering a listening ear to frustrations will be really helpful.
3. Learn to prepare some favourite special dishes. If you are having a person with maple syrup urine disease over for a meal, ask them what they would like to eat and offer low-protein snack options such as:
 - soda pop
 - fruit juices
 - vegetable sticks with non-dairy dips
 - fruit plate

Resources

Maple Syrup Urine Disease family support group
www.msud-support.org
