

# MAPLE SYRUP URINE DISEASE

## FACT SHEET



*This fact sheet is for educational purposes only. An experienced healthcare professional should be consulted for the management of maple syrup urine disease.*

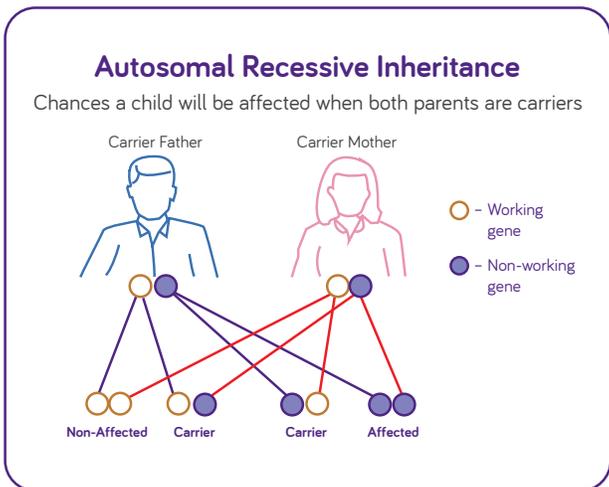
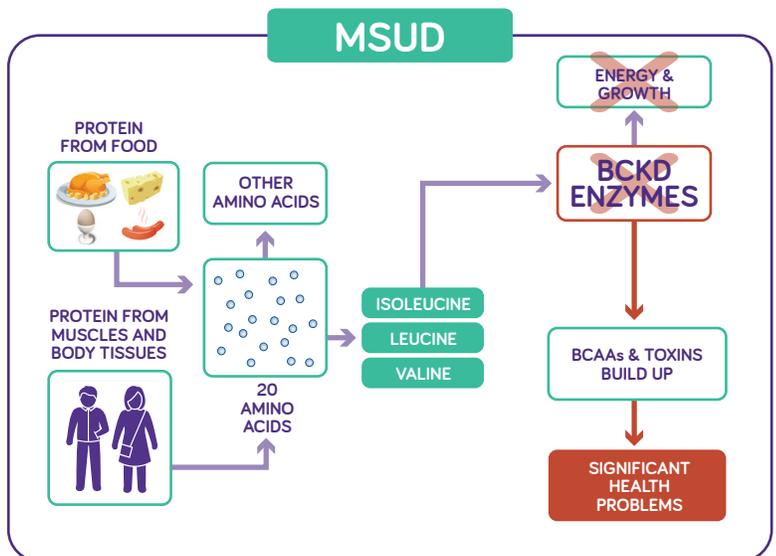
### What is Maple Syrup Urine Disease?

Maple syrup urine disease (MSUD) is an inherited, genetic disorder caused by a defect in three enzymes that help break down protein from food. The official name for MSUD is branched-chain alpha-ketoacid dehydrogenase deficiency, but it is called MSUD because the urine of untreated infants with this disorder can smell sweet like maple syrup.

Protein is made up of smaller units called amino acids. Amino acids are important to build muscle and other body tissues for normal growth. Any extra amino acids are normally broken down to produce energy. In MSUD, three of the amino acids cannot be broken down in the body. These amino acids are leucine, isoleucine and valine, which are called branched-chain amino acids (BCAAs). When someone with MSUD eats too much protein from food, does not get enough calories, or becomes ill, blood levels of the BCAAs increase and so do their byproducts, called ketoacids.

High levels of BCAAs, especially leucine and its ketoacid, can cause many health problems including poor feeding, vomiting, severe weakness (lethargy), seizures and coma. If not treated aggressively, MSUD can be fatal. Long term problems can include poor growth, slow development and learning problems.

Finding out that your child has MSUD can be overwhelming. Fortunately, nutrition management is available to help prevent many of these problems. Take comfort in knowing that your metabolic healthcare team can provide you with the best advice to help manage MSUD.



### Incidence, Genetics and Newborn Screening

MSUD is a rare disorder that occurs in about 1 in every 185,000 births. It occurs more often in the Mennonite community, affecting about 1 in every 380 infants.

MSUD is inherited as a “recessive disorder.” In recessive disorders, both the mother and father are “carriers,” and they do not have any symptoms of the disorder (see diagram). With each pregnancy there is a 1 in 4 (25%) chance that the infant will be affected. A genetic counselor can help explain the inheritance of MSUD and the risks to future infants.

MSUD can be screened for at birth through a simple blood test. For details on newborn screening (NBS) in the US, visit [www.newbornscreening.hrsa.gov/your-state](http://www.newbornscreening.hrsa.gov/your-state); for information on NBS in Canada, visit [www.raredisorders.ca](http://www.raredisorders.ca).

## Management of Maple Syrup Urine Disease

There is no cure for MSUD, but it can be managed with a special diet. The purpose of the diet is to provide just enough leucine, isoleucine and valine for proper growth, but avoid excess intake of these amino acids. Typically, special medical formulas and a low protein diet are necessary. It is important that an individual with MSUD continues management for life.

A medical formula is a very important part of the diet for MSUD. The protein source in MSUD medical formulas is individual amino acids, but the BCAAs are left out. This allows a person with MSUD to get enough protein without the parts of protein that can be harmful. Medical formulas may also provide calories, vitamins and minerals the body needs for normal growth. For some, extra isoleucine and valine may be added to the formula to prevent deficiencies of these two amino acids.

To provide just enough BCAAs that a person with MSUD needs, a limited amount of natural protein-containing foods are allowed in the diet. For infants, breast milk or regular infant formula is given in precise amounts. As the baby grows and can eat solid foods, the breast milk or regular infant formula will be removed from the diet and the BCAAs will come from foods instead.



For the MSUD diet, all high protein foods are avoided, as eating too much high protein food increases blood levels of BCAAs. High protein foods include milk and dairy products, meat, poultry, fish, eggs, beans, nuts and peanut butter. On the other hand, vegetables and fruit are allowed. Regular breads and pastas may be allowed in small amounts, but special low protein versions of these foods are often used instead to allow for more choices in the diet and less concern for eating too much protein. Foods provided in the MSUD diet may be weighed or measured to ensure that excess protein is not eaten. Typically, the amount of leucine in the diet is counted. A metabolic dietitian works closely with people with MSUD to prescribe the best diet plan and help with needed changes in the future.

Nutricia North America provides a range of MSUD formulas for MSUD. Please contact us for more information. Your dietitian will help you decide which products are best.

## During Illness

During illness or after an injury, the body increases the breakdown of protein from muscles and organs. This increases the risk of high levels of BCAAs and ketoacids in the blood, which can lead to serious medical problems. Early signs of high BCAA levels include vomiting, excessive sleepiness, coordination problems and/or changes in mental status. During any illness, it is very important to notify your metabolic clinic immediately. Often, the diet is adjusted to decrease leucine intake and increase calories. This can help slow the breakdown of protein from the body. Your clinic will give you an emergency letter – if you notice signs of high BCAA levels, take this letter to the emergency room. During an illness, a hospital stay may be needed.

## Resources

- Screening Technologies and Research in Genetics (STAR-G): [newbornscreening.info](http://newbornscreening.info)
- Newborn Screening in Your State (US): [newbornscreening.hrsa.gov/your-state](http://newbornscreening.hrsa.gov/your-state)
- MSUD Family Support Network: [msud-support.org](http://msud-support.org)
- Canadian Organization for Rare Disorders: [raredisorders.ca](http://raredisorders.ca)

*Nutricia North America would like to thank Sandy van Calcar, PhD, RD, Oregon Health & Science University for her consultation.*

Questions about MSUD  
Formula Coverage?



**NUTRICIA  
NAVIGATOR**

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For more information or product samples, please visit [NutriciaMetabolics.com](http://NutriciaMetabolics.com)  
or call US: **(800) 365-7354**

All products shown are medical foods in the US and foods for special dietary use in Canada for the dietary management of maple syrup urine disease (MSUD) and must be used under medical supervision.