

This fact sheet is for educational purposes only. An experienced healthcare professional should be consulted for the management of methylmalonic or propionic acidemia.

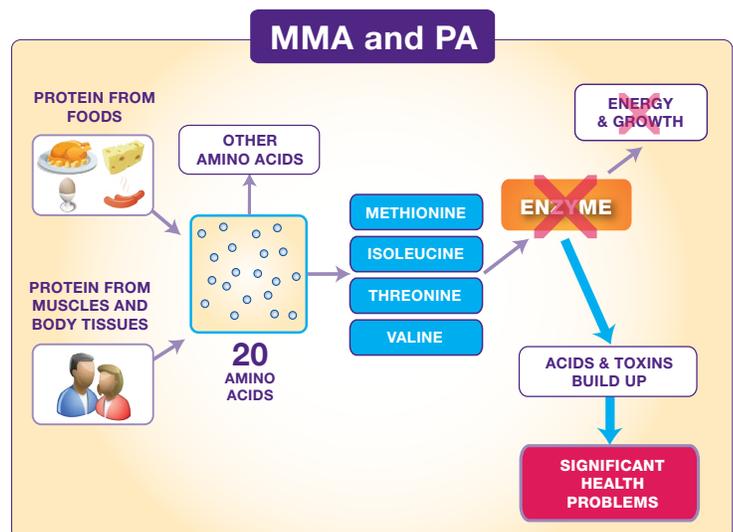
What are Methylmalonic Acidemia and Propionic Acidemia?

Methylmalonic acidemia (MMA) and propionic acidemia (PA) are inherited, genetic disorders. Both are caused by defects in enzymes that are needed to break down protein. Protein is made up of amino acids, which are important to build muscle and other body tissues for normal growth. Any extra amino acids are normally broken down to produce energy. In MMA and PA, four of the amino acids cannot be broken down but instead produce toxic acids in the body. These amino acids are **methionine, isoleucine, threonine** and **valine**, which are called “propionic amino acids.”

When someone with MMA or PA either eats too much protein from food, does not get enough calories or becomes ill, blood levels of the propionic amino acids increase and so do their toxic byproducts. The toxic acids are methylmalonic acid, which is present in MMA, and propionic acid, which is present in both MMA and PA. Bacteria in the digestive system also produce propionic acid. Both toxic acids can cause serious and life-threatening health problems. The enzyme that is missing or not working properly in MMA is called methylmalonyl-CoA mutase, and in PA it is called propionyl-CoA carboxylase.

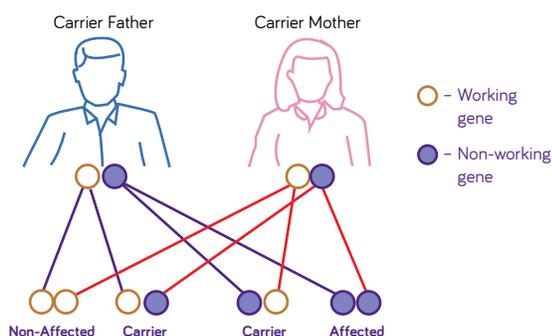
High blood levels of propionic amino acids and their related toxic acids can cause many health problems including poor feeding, vomiting, severe weakness (lethargy), seizures and coma. If not treated aggressively, both disorders can be life-threatening. Long term problems can include poor growth, abnormal movements (ataxia) and learning problems.

Finding out that your child is diagnosed with MMA or PA 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 the disorder.



Autosomal Recessive Inheritance

Chances for each child when both parents are carriers



Incidence, Genetics and Newborn Screening

MMA and PA are rare disorders that occur in about 1 in every 80,000 to 100,000 births.

Both MMA or PA are inherited as “recessive disorders.” 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 the disorder and the risks to future infants.

Both MMA and PA can be screened for at birth through a simple blood test. For details on newborn screening (NBS) in the US, visit <https://newbornscreening.hrsa.gov/your-state>; for information on NBS in Canada, visit <https://www.raredisorders.ca/>.

Management of Methylmalonic Acidemia and Propionic Acidemia

There is no cure for MMA or PA, but they can be managed with a modified diet, medication, and special medical formulas specifically designed for persons with MMA or PA. The modified diet for these disorders is low in propiogenic amino acids. It is important that an individual with MMA or PA continues with management as directed by their healthcare professional.

A medical formula may be an important part of the diet for MMA and PA. The protein source in MMA and PA medical formulas is individual amino acids, but the four propiogenic amino acids are either not included or two of the four are included at small amounts. This allows a person with MMA or PA to get enough protein without the parts of the protein that can be harmful. Medical formulas may also provide calories, vitamins and minerals the body needs for normal growth.

To provide just enough of the propiogenic amino acids that a person with MMA or PA 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 propiogenic amino acids will come from foods instead. Some individuals with MMA or PA may require a feeding tube. When a feeding tube is used all of the nutrients may come from MMA/PA formula and either breast milk or regular formula.

Since all foods with protein contain the four propiogenic amino acids, individuals with MMA or PA must limit their intake of foods high in protein. These include milk and dairy products, meat, poultry, fish, eggs, beans, nuts and peanut butter. Regular breads and pastas may be allowed in certain amounts, but special low protein versions of these foods are often used instead. Vegetables and fruits are allowed. Foods may be weighed or measured to ensure that excess protein is not eaten. Typically, the amount of protein in the diet is counted. A metabolic dietitian will work closely with people with MMA or PA to prescribe the best diet plan and help with needed changes in the future.

Foods Low in Protein



Nutricia North America provides a range of medical formulas as well as low protein foods. Please contact us for more information. Your dietitian will help you decide which products are best.

Some people with MMA have a form of the disorder that is called “vitamin B₁₂ responsive.” For these people, high doses of vitamin B₁₂ may be given as an injection to help reduce levels of methylmalonic acid in the blood.

A drug called L-carnitine may be prescribed to those with MMA and PA. L-carnitine binds to propionic acid or methylmalonic acid and thus reduces the toxicity of these acids. Your metabolic doctor will decide if L-carnitine or other drugs are needed. For some patients, a liver transplant may be an option.

During Illness

During an illness or after an injury, the body increases the breakdown of protein stores. For those with MMA or PA, this can increase the level of propionic or methylmalonic acid in the blood. This can lead to serious medical problems. Early signs of illness include vomiting, excessive sleepiness, coordination problems and/or changes in mental status. During any illness or after an injury, it is very important to notify your metabolic clinic immediately. Often, the diet is changed to decrease protein and increase calories. This can help slow down the breakdown of protein stores. Your clinic will give you an emergency letter – if you notice signs of high propionic or methylmalonic acid levels, take this letter to the emergency room. During serious illness, a hospital stay may be needed.

Resources

- Screening Technologies and Research in Genetics (STAR-G): <http://www.newbornscreening.info/>
- Newborn Screening in Your State (US): <https://newbornscreening.hrsa.gov/your-state>
- Organic Acidemia Association: <https://www.oaanews.org/>
- Propionic Acidemia Foundation: <http://www.pafoundation.com/>
- Canadian Organization for Rare Disorders: <https://www.raredisorders.ca/>

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

Questions about MMA/PA formula coverage?



**NUTRICIA
NAVIGATOR**

📞 800-365-7354

✉ NutriciaNavigator@Nutricia.com

For more information or product samples, please visit NutriciaMetabolics.com or call US: (800) 365-7354 Canada: (877) 636-2283

All products shown are medical foods in the US and foods for special dietary use in Canada for the dietary management of methylmalonic acidemia (MMA) and propionic acidemia (PA) and must be used under medical supervision.

Formula for MMA/PA



*Only available in US