An experienced healthcare professional should be consulted for the management of PA or MMA.

**What Are Propionic Acidemia And Methylmalonic Acidemia?**

Propionic acidemia (PA) and methylmalonic acidemia (MMA) 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 PA and MMA, 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 “propiogenic amino acids.”

When someone with PA or MMA either eats too much food protein, eats too few calories or becomes ill, blood levels of the propiogenic amino acids increase and so do toxic byproducts of these amino acids. The toxic acids are propionic acid, which is present in PA, and methylmalonic acid, which is present in MMA. 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 PA is called propionyl-CoA carboxylase, and in MMA it is called methylmalonyl CoA mutase.

High blood levels of propiogenic 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 (also called ataxia) and learning problems.

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

**Incidence, Genetics and Newborn Screening**

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

Both PA and MMA are “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 PA and MMA can be screened for at birth through a simple blood test. For more details on newborn screening in your state or province, please visit http://genes-r-us.uthscsa.edu.
Management of Propionic Acidemia and Methylmalonic Acidemia

There is no cure for PA or MMA, but they can be managed with a modified diet, medication, and special medical formulas specifically designed for persons with PA or MMA. The modified diet for these disorders is low in propiogenic amino acids. It is important that an individual with PA or MMA remains on treatment for life.

A medical formula is a very important part of the diet for PA and MMA. The protein source in PA and MMA 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 PA or MMA 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 PA or MMA 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 PA or MMA may require a feeding tube. When a feeding tube is used all of the nutrients come from PA and MMA formula and either breast milk or regular formula.

Since all foods with protein contain the four propiogenic amino acids, individuals with PA or MMA 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 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. Vegetables and fruit are allowed. Foods are usually weighed or measured to ensure that extra protein is not eaten. Typically, the amount of protein in the diet is counted. A metabolic dietitian will work closely with people with PA or MMA to prescribe the best diet plan and help with needed changes in the future.

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 B_{12} responsive methylmalonic acidemia.” For these people, high doses of vitamin B_{12} 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 PA and MMA. 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.

If You or Your Child Is Sick

During an illness or after an injury, the body increases the breakdown of protein stores. For those with PA or MMA, 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 a 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

- Star-G website: http://www.newbornscreening.info
- Family organizations: Organic Acidemia Association (www.oaanews.org) and Propionic Acidemia Foundation (www.pafoundation.com)

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For more information about Nutricia products and great recipes, visit www.myspecialdiet.com