

## UNDERSTANDING PROPIONIC ACIDEMIA

### What is propionic acidemia?

Propionic acidemia (PRO-PEE-ON-IC ASID-EE-MEEA) is genetic disorder that affects how protein is broken down in the body. It is a **metabolic** disorder. There are about 1 in 50 000 to 1 in 100 000 people with propionic acidemia worldwide.

**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 propionic acidemia are unable to properly break down some of these amino acids because they are missing an **enzyme** in their cells. Some people with propionic acidemia have small amounts of this enzyme and some have none at all. Those with some of the enzyme respond to biotin supplements in the diet (see medications section for more information on biotin). Those with no working enzyme do not respond to biotin.



**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 that has been eaten is like the delivery. In propionic acidemia, when the enzyme is missing, protein cannot be fully broken down for use by the body. It is only broken down to the 'acid' stage. This creates problems for the person with propionic acidemia. These acids build up in the blood, fluid around the brain and spinal cord, and urine. The name "propionic acidemia" means propionic acid in the blood. The acid is like the delivery person's

packages. It 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 acid is not removed quickly, the person may have seizures or may go into a coma.

In untreated propionic acidemia, the balance is upset. There is too much 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

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

### **How is propionic acidemia treated?**

The treatment for propionic acidemia is aimed at preventing the toxic effects of elevated propionic acid. Therapy needs to begin before complications occur because, once they develop, they are irreversible. Examples of this are bone problems and mental retardation from swelling around the brain. Treatment must be started as soon as the disorder is diagnosed.

Treatment for propionic acidemia typically includes the following:

## DIET



People who have propionic acidemia and who have *no* working enzyme (they do not respond to biotin) 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 high protein foods like meat and dairy products. Examples of low protein foods are fruits, vegetables and starches. These foods provide calories without loading the body with excessive protein.

**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 propionic acidemia to make adjustments to diet. This ensures that adequate calories and nutrients are being provided. It is often important for the person to keep a record of what they eat on a daily basis.

The amount of food and drink the person can safely consume is calculated by the dietitian. Foods may need to be carefully measured to control the amount of protein consumed.

Examples of foods that must be eliminated (or significantly limited) in propionic acidemia 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. In order to prevent complications, the person must adhere to this diet for life.

### **The balancing act**

The challenge in treating propionic acidemia is to provide enough protein to meet body requirements without overloading the person with protein waste (acid). 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.

## MEDICATION



### 1. Flagyl

Flagyl (or metronidazole) helps to decrease the production of acids by bacteria in the intestines. This helps to decrease the overall acid levels in the blood. Flagyl is taken in pill form and is available by prescription only.

### 2. Biotin

Biotin is a naturally occurring substance in the body. It acts as a helper for the enzyme that breaks down propionic acid. Some people with propionic acidemia respond well to biotin therapy. Biotin is available by prescription in capsule form.

### 3. Carnitine

Carnitine is prescribed to people with propionic acidemia. Taking carnitine is important for people with the disorder for two reasons:

- People with propionic acidemia don't have enough carnitine in their bodies
- the low protein diet doesn't provide carnitine from natural sources

Carnitine also helps rid the body of toxic metabolites that build up in people with propionic acidemia. Carnitine is available in liquid or pill form.

## How do people get propionic acidemia?

Propionic acidemia 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 propionic acidemia, 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 propionic acidemia but are not affected. There is a 1 in 4, or 25% chance that two carriers of the gene will have a baby with propionic acidemia. Boys and girls are equally affected.

With each pregnancy, there is the **same** 25% chance of having a child with propionic acidemia. In some families, there may be only one child with propionic acidemia, 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 propionic acidemia 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 propionic acidemia 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 propionic acidemia 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
  - candies

## Resources

Organic Acidemia Association  
13210 35<sup>th</sup> Avenue North  
Plymouth, MN 55441  
Phone: (763) 559-1797  
Fax: (763) 694-0017  
Email: [OAAnews@aol.com](mailto:OAAnews@aol.com)  
Internet: [www.oaaneews.org](http://www.oaaneews.org)

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