



PROPIONIC ACIDEMIA^(PA)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called propionic acidemia.

A positive newborn screen does not mean your baby has propionic acidemia, but it does mean your baby needs more testing to know for sure.

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing.

What is propionic acidemia?

Propionic acidemia affects an enzyme needed to break down certain proteins and fats in the food we eat, so they can be used for energy and growth. In propionic acidemia, the enzyme used to break down these proteins and fats is missing or not working properly.

A person with propionic acidemia doesn't have enough enzyme to break down protein which can cause harmful toxins to build up in the body.

Propionic acidemia is a genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause propionic acidemia. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can propionic acidemia cause?

Propionic acidemia is different for each child. Some children with propionic acidemia have few health problems, while other children may have a severe form of propionic acidemia with serious complications.

If propionic acidemia is not treated, a child might develop:

- Poor feeding
- Vomiting
- Sleepiness (lethargy)
- Poor muscle tone
- Serious illness (metabolic crises)
- Seizures
- Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for propionic acidemia?

Propionic acidemia can be treated. Treatment is life-long and can include:

- Low protein diet – a dietitian will help you set up the best diet for your child.
- Medications to help the body get rid of propionic acid and make energy.

Children with propionic acidemia should see their regular doctor, a doctor who specializes in propionic acidemia, and a dietitian.

Prompt and careful treatment helps children with propionic acidemia live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening

Nurse Consultant

Toll-free: 1-866-673-9939

newbornscreening@michigan.gov

Michigan Metabolic Coordinating Centers

Children's Hospital of Michigan: 1-313-832-9343

Michigan Medicine: 1-734-764-0579

Children's Special Health Care Services

Toll-free: 1-800-359-3722



Phone 1-866-673-9939

www.michigan.gov/newbornscreening

(Based on and printed with permission from Minnesota Newborn Screening)