Propionic Acidemia

Other names: Propionyl-CoA carboxylase deficiency, PCC deficiency **Gene involved:** *PCCB*

What is Propionic Acidemia?

Propionic acidemia (PA) is an inherited condition that affects the way our bodies use energy from protein. Normally, as we make energy from protein, our bodies clear the chemicals we don't need. One of these chemicals is called propionic acid. People with PA cannot clear propionic acid from their bodies. This can lead to problems with their heart and brain. Children are born with PA and do not grow out of it.

What are the early signs and symptoms?

The first signs of PA may occur in newborns, children or even adults. Some babies may develop symptoms within the first few days of life, including:

- Poor muscle tone
- Poor feeding
- Seizures
- Coma
- Most babies will look healthy, but childhood illnesses and flus can suddenly worsen PA symptoms

What Happens as Children Get Older?

- Some children have learning difficulties, physical disabilities or behavior problems
- Some people may develop heart problems (cardiomyopathy) and can have heart rhythm problems
- Infections and heavy exercise can cause the body to start breaking down its protein stores, leading to a 'PA crisis' with vomiting, seizures and coma

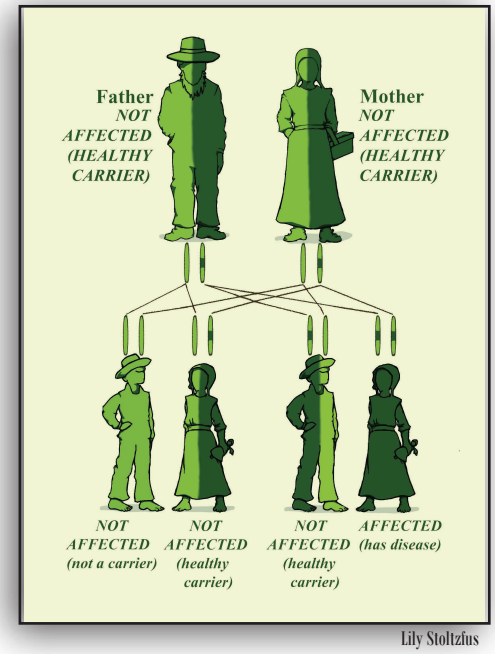
Management

The main principles of treatment are:

- Limiting protein in food
- Avoiding fasting
- Close communication with the doctor when sick
- Using supplements to clear propionic acid
- Expectant mothers with PA should be closely monitored during pregnancy

Diagnosis

A gene test is required to make the diagnosis. To arrange this, please see the contact information below.



Lily Stoltzfus

Autosomal Recessive Inheritance

Genes are instructions for growth and development. We have two copies of each gene. One copy is inherited from our mother, and one copy is inherited from our father. Children with PA have a spelling change in BOTH copies of their *PCCB* genes. This means they have no working copy of the *PCCB* instruction.

People with one normal copy of the *PCCB* gene and one misspelled copy are called "healthy carriers".

When two healthy carrier parents have a child, there is a:

- 1 in 4 chance the child will have PA, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *PCCB* gene and be neither affected nor be a carrier

This informational pamphlet is a project of New Leaf Center - a member of the Plain Community Health Consortium (PCHC)

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Center for Special Children (La Farge, WI) **608-625-4039**
Central Pennsylvania Clinic (Belleville, PA) **717-935-2065**
Clinic for Special Children (Strasburg, PA) **717-687-9407**
DDC Clinic (Middlefield, OH) **440-632-1668**
Midwest Genetics Network (Okemos, MI) **517-324-8300**
The Community Health Clinic (Topeka, IN) **260-593-0108**
UPMC Children's Hospital of Pittsburgh (Pittsburgh, PA) **412-692-5070**
WeCare Clinic - Medical Care for Special Needs (Pembroke, KY) **270-962-7383**