

Frequently asked questions about pyruvate dehydrogenase complex deficiency

What is pyruvate dehydrogenase complex deficiency (PDCD)?

PDCD is a problem within the energy making cells of the brain called mitochondria. The mitochondria make energy for other cells to grow and develop. Mitochondria need many enzymes in order to work properly. In this disorder, an enzyme is not available in adequate amounts to help make energy.

Is it contagious?

PDCD is not contagious. It is passed on through recessive genes. Genes come in pairs. Recessive inheritance means both genes in a pair must be defective to cause a condition. People with only one defective gene in the pair are considered carriers. However, they can pass the abnormal gene to their children.

How is it diagnosed?

A blood level of lactate and pyruvate will be elevated (although these may be elevated in other conditions). Blood and urine levels of certain proteins called amino acids may also be elevated. Another compound in the blood called ammonia may be elevated, especially during illness. Imaging studies such as an MRI can show structure problems in the brain.

What are the symptoms?

Symptoms usually start in infancy but may start at birth or in later childhood. These symptoms may include developmental delay, poor muscle tone, abnormal eye movements, difficulty walking and seizures.

Is this syndrome common?

PDH is considered a rare condition but one of the most common neurodegenerative disorders that is related to a problem in the mitochondria's ability to make energy.

What is the treatment?

According to a recent publication in 2008 by a group of medical experts, the Ketogenic Diet is the treatment of choice for PDCD. More information about this diet can be found under the FAQ link of our web-site: charliefoundation.org.

What benefits can be expected if the ketogenic diet is started?

Increased alertness and ability to learn are usually experienced soon after starting the ketogenic diet. Detecting this disorder early is the key to helping the brain receive the energy it needs to grow and to develop.