Propionic Acidemia

What is Propionic Acidemia?
Propionic Acidemia, also known as Propionyl-CoA Carboxylase Deficiency, is an inherited disorder in which the body is unable to process certain proteins and lipids properly. This condition leads to abnormal buildup and levels of organic acids in the blood, urine, and tissues, which can be toxic and cause serious health problems. The condition appears usually in early infancy, is characterized by poor feeding, vomiting, weak muscle tone, and lethargy.

How You Can Help:
If able, donate! Rare diseases are severely underfunded and require financial support to fund research and treatments.

Have conversations! Raising awareness is critical. As this is a rare disease, not many people know what Organic Acidemia is, much less propionic acidemia. Share this with your friends, family, health care providers, and government.

Join the cause! After all we are stronger in numbers.

For more information, go to www.oaanews.org

Causes:
Mutations in PCCA and PCCB genes cause propionic acidemia. These genes provide instructions for making an enzyme called propionyl-CoA carboxylase. This enzyme helps process several amino acids, which are the building blocks of proteins. It also helps break down certain fats and cholesterol. Mutations result in propionyl-CoA build up to toxic levels, damaging the brain and nervous system. This disease is inherited in an recessive autosomal pattern, meaning both parents must carry a copy of the mutated gene.

SOURCE: MedlinePlus

Barriers to Care:
- As PA is a rare disease there is a lack of knowledge within the medical community that makes the disease hard to diagnose and treat.
- Misdiagnoses are common, and setback the process of receiving proper care.
- PA affects 1 in 100,000-250,000 people around the world. This small population size effects pharmaceutical decisions.
- Costly care and insurance.
- Only available treatment are protein formulas.

SOURCE: National Organization for Rare Disorders
An Interview with Dr. Venditti

Dr. Venditti has been doing research on the Organic Acidemias since the late 1990s. His passion for the diseases started while observing the expansion of newborn screenings. After seeing the importance of finding and treating patients as early as possible to improve the outcomes of their conditions, Dr. Venditti’s interest in the Organic Acidemias began.
Methylmalonic Acidemia: An Interview with Sienna

Sienna’s Thoughts:

- Individuals and families with genetic disorders should “know that you’re not alone”
- OAA and connected programs “provide great support”
- Very hopeful in progress in gene therapy towards finding a more effective treatment
- High prices of medication for MMA and connected maladies are challenging for many

For more go to www.oaanews.org

What is Methylmalonic Acidemia (MMA)?:

- A genetic Disorder which makes it difficult to break down certain fats and proteins
- Glycine and methylmalonic acid build up in blood and urine
- There can be particular difficulty processing the amino acids isoleucine, valine, methionine, and threonine
- Individuals may exhibit breathing, motor function, vision, and cognitive difficulties in addition to lethargy and a lack of appetite

Source: National Institute of Health and Genetic, Rare Disease Information Center
What are Organic Acidemias?

Organic Acidemias are a group of rare diseases that are categorized by an enzyme deficiency that causes the buildup of organic acids in the blood and urine.

There are 21 different types of Organic Acidemias. Some of the most common include:
- Methylmalonic Acidemia
- Propionic Acidemia
- Isovaleric Acidemia

Treatment Includes:
- A specific diet or a protein formula
- Supplements

Source: NORD, OPHC/Children's Hospital of Wisconsin

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