

Methylmalonic Acidemias (MMA) & Propionic Acidemia (PA)

General Overview

Q. What are MMA/PA?

A. MMA/PA are disorders that affect the way the body processes protein. They are treatable, but can cause life-threatening illness.

Q. Is there only one form of MMA/PA?

A. No. MMA/PA includes at least five different disorders. Some show up very early, within the first days or weeks of life, and can be life-threatening. Others show up later but usually within the first year of life. All can cause severe physical and mental damage.

Q. How does the body normally process proteins?

A. Proteins are made up of amino acids. These amino acids are normally used to make new proteins or broken down further by enzymes to make energy.

Q. What happens to proteins in a child with MMA/PA?

A. In a child with MMA/PA, some amino acids are not broken down properly, and by-products, including methylmalonic or propionic acid, build up in the blood and body.

Q. What is the treatment for MMA/PA?

A. MMA/PA are treated with a special diet that is low in the amino acids isoleucine, valine, methionine and threonine. Treatment should begin shortly after birth, and is life-long. Since protein is essential for growth and development, the child must continue on a special formula that provides protein and essential nutrients, but contains little or no isoleucine, valine, methionine and threonine. Some doctors may also prescribe other medicines or dietary supplements. People with MMA/PA require treatment through a specialty clinic with experience in treating these disorders.

Q. Why would a child have MMA/PA?

A. MMA/PA are inherited disorders. They result when a baby receives a double-dose of the same non-working gene (one from each parent) involved in breaking down valine, isoleucine, methionine, and threonine. For more information about this, contact your health care provider or a genetic counselor.

Q. How common is MMA/PA?

A. About one in every 57,000 babies in the United States is born with MMA or PA. However, parents who have a child with MMA/PA each carry one copy of the non-working gene. That means, with each pregnancy, each one will have a one in four chance of having MMA/PA.

For more information about MMA/PA, please see the Disorders section of our website: www.doh.wa.gov/nbs.

