Commonly asked questions

1. What is PA or MMA?

PA is also known as propionic acidemia. MMA is also known as methylmalonic acidemia. They are organic acid disorders and represent defects in the metabolism of certain essential amino acids. The inability to completely metabolise these amino acids leads to a build up of toxic intermediate chemicals. This is often excacerbated at times when the body is stressed(e.g. fasting, operations or infections). During these times the body breaks down its own proteins to supply needed energy and as a result, the amino acids are metabolized into the toxic intermediates.

2. How and when will we know if my baby has PA or MMA?

If your baby's newborn screening result showed a markedly elevated C3 levels, he or she probably has PA or MMA. The newborn screening test will be repeated and additional tests will be undertaken to help determine whether or not your baby has PA or MMA. Typically, the results of these tests take up to 4 days to come back. Depending on the test results, additional testing can take a variable amount of time to confirm the diagnosis. In a very small minority of cases it can be difficult to determine whether or not a child is affected.

3. How did my baby get this?

PA and MMA are autosomal recessive genetic disorders. This means that your baby has two abnormal genes, one from the mother and one from the father. Having only one mutated gene (a carrier) does not affect a person at all.

4. What does it mean for my child?

If your baby has PA or MMA, he or she will have to have a special protein restricted diet. Most children with these conditions also take carnitine, a mild supplemental medicine. If your child becomes ill, it may well be necessary early in the illness (i.e. when it might be considered mild), to further restrict the protein intake for a short period of time or even to provide extra energy in the form of glucose through addition to food or, if necessary, by intravenous infusion. By treating your baby this way it is possible to generally prevent the worst effects of these conditions. However, babies and children with PA or MMA are at risk from serious effects such as mental retardation , loss of control of movement or even death if allowed to get sick throughout childhood. Therefore, it is important to maintain vigilance, consider every illness seriously and hospitalize for specialized treatment early. Some children, despite the best treatment and care possible, will still have some delay though this will be significantly less than if your child is not treated as described above. Children with PA also tend to have feeding difficulties up to the early school years when this problem typically abates.

5. What is the treatment? Does it work? Is the diet difficult to do/expensive?

PA or MMA is primarily treated by a protein restricted diet with supplemental amino acid formula. The special formula which will keep your child well is typically ordered through your metabolic clinic where the metabolic nutritionist will ensure that you are confident in preparing it. The formula can be expensive; however, your metabolic clinic will assist you in obtaining it through your health care provider or state agency.

6. What about my other children/future children?

As PA and MMA ire inherited conditions it is essential to have your other children tested. Children from the same father and mother as the affected infant have a 1 in 4 (25%) chance of having the same condition. Your other children can appear healthy and still

have the disease. If they have PA or MMA, successfully having weathered illnesses in the past is no guarantee that an illness in the future will not have serious consequences. Since there is a risk for having a future child with PA or MMA it is important to let your obstetrician and pediatrician know that you have a child with PA or MMA if you are planning future pregnancies so that they may discuss the options with you and prepare accordingly.

acylcarnitines and plasma amino acids. See Acute illness protocol, PA or MMA.

4. Enzyme assay. Go to genetests

Propionyl-CoA carboxylase and methylmalonyl-CoA mutase enzymatic activity can be measured in cultured fibroblast cells (e.g. from a skin biopsy).

5. Molecular testing

Genotyping is available for both PA and MMA. Go to genetests