

## Commonly asked questions

### **1. What is MSUD?**

MSUD, also known as maple syrup urine disease, is an organic acid disorder caused by a defect in the metabolism of a specific group of amino acid called branched chain amino acids. The inability to completely metabolize these amino acids leads to a toxic build up of these and related acids in the body. This is often exacerbated when the body is stressed (e.g. newborn period, fasting, operations or infections). During these times the body breaks down its own proteins releasing these amino acids to supply needed energy and as a result, these toxins accumulate.

### **2. How and when will we know if my baby has MSUD?**

If your baby's newborn screening result showed a markedly elevated leucine level, especially if valine (another branched chain amino acid) was also elevated, he or she probably has MSUD. Hydroxyprolinemia, which produces no harm, could also be the diagnosis, especially if only leucine seems to be increased. The newborn screening test will be repeated and additional tests will be undertaken to help determine whether or not your baby has MSUD. The results of these tests may 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. Very rarely, it can be difficult to determine whether or not your baby is affected.

### **3. How did my baby get this?**

MSUD is an autosomal recessive genetic disorder. This means that if your baby has MSUD, he/she has two abnormal mutated genes, one from the mother and one from the father. Each of you will have one mutated gene (a carrier). Being a carrier does not affect a person at all.

### **4. What does it mean for my child?**

If your baby has MSUD, he or she will have to be on a special protein restricted diet, and also take a special formula to ensure that the diet is adequate and balanced. 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 additional sugar 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 MSUD are at risk for learning disabilities, mental retardation, or even death if allowed to get sick and not receive the necessary immediate treatment. Therefore, it is important to maintain vigilance and consider every illness seriously. 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.

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

MSUD is primarily treated by a protein-restricted diet and special 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. Your metabolic clinic will assist you in obtaining the formula through your health care provider or state agency.

## **6. What about my other children/future children?**

As MSUD is an inherited condition 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 disorder. If they have MSUD, successfully having weathered illnesses in the past is no guarantee that an illness in the future will not have serious consequences. If they were not screened for MSUD when they were babies, they should be tested.

Since there is a risk (1 in 4 or 25% in each pregnancy) for having a future child with MSUD it is important to let your obstetrician and pediatrician know that you have a child with MSUD if you are planning future pregnancies so that they may discuss the options with you and prepare accordingly.