Why does my baby have to have more tests?

At about 2 days of age your baby had a heel prick blood test where drops of blood were soaked in to a special card and sent to the newborn screening laboratory. The results of this screening showed a significantly elevated level of a substance called leucine. This strongly suggests your baby has a disorder called Maple Syrup Urine Disease. Your baby needs to have urgent blood tests and a urine test to confirm the diagnosis. S/he also needs to be checked by a paediatrician.

What is Maple Syrup Urine Disease?

Maple Syrup Urine Disease or MSUD (also called branched-chain ketonuria), is a genetic disorder. The name comes from the characteristic odour of the urine of affected infants. It is a very rare condition with only about one in every 100,000 to 200,000 babies born having this disorder. People with MSUD have a deficiency of an important enzyme that helps to break down some amino acids. Amino acids are the building blocks of protein. Because of this enzyme deficiency, three amino acids, leucine, isoleucine and valine build up in blood and urine and presumably throughout the body. An accumulation of these amino acids causes severe illness unless effective treatment is given.

What are the symptoms of MSUD and how is it diagnosed?

With the usual form of MSUD, low muscle tone, lethargy (sleepiness), poor feeding and low blood sugar levels develop in the first week or two in an infant who seemed normal at birth. Unless the baby is treated quickly, seizures and coma may develop, and without treatment these babies would die in the first few weeks of life. The diagnosis can sometimes be suspected from the characteristic odour of the urine. It is confirmed by the abnormal pattern of amino acids and keto acids in blood and urine.

How is MSUD treated?

Treatment is by careful dietary control of leucine, isoleucine, and valine. This means a very low protein diet, and a special supplement containing all the amino acids the body needs, except leucine, isoleucine and valine. With an early introduction of this diet and careful monitoring of blood amino acid levels most children have developed well.

A few patients have been diagnosed with milder variants of maple syrup urine disease. Some of these patients may respond to thiamine (one of the B group vitamins), but this treatment is not useful in the usual form of MSUD.

How did my baby get MSUD?

MSUD is a genetic disorder that is inherited from both parents by what is called recessive inheritance. All cells in the body carry two copies of each gene, one inherited from each parent. In all of us, there are mistakes or “mutations” in a few of the genes. For a person to have MSUD they must have inherited two faulty mutations of the MSUD gene, one from each parent. There is nothing you could have done to prevent this from happening.