



INFORMATION FOR PARENTS/CARERS 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. The disease is transmitted as a rare autosomal recessive trait*. Only about one in every 100,000 to 200,000 babies born have this disorder. People with MSUD have a deficiency of an important enzyme**. Because of this enzyme deficiency, three amino acids, leucine, isoleucine and valine accumulate in excess in blood and urine and presumably throughout the body, and cause toxic effects. Aminoacids are the building blocks of protein.

With the usual form of MSUD, low muscle tone, lethargy, poor feeding and low blood sugar levels develop in the first week 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 and is confirmed by the abnormal pattern of amino acids and keto acids in blood and urine.

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 aminoacids the body needs, *except* leucine, isoleucine and valine. With early institution of dietary control and careful monitoring of blood aminoacid levels the results have been gratifying, and the children have developed well.

A few patients have been described with milder variants of maple syrup urine disease. These mild variant patients may respond to the B vitamin thiamine, but this treatment is not useful in the usual form of MSUD.

***Recessive inheritance:** each cell in the body carries 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 recessive gene mutation to result in a particular disorder, both copies of the gene must carry a mutation.

****Enzyme:** an enzyme is a protein made in the body, which helps a chemical reaction to proceed. Each of the chemical reactions or steps, in the body’s metabolism needs a specific enzyme to function. Enzymes are needed, for example, in all steps in the breakdown of the food we eat, so that our bodies can use it.

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For further information please contact your paediatrician or the NSW Newborn Screening Programme

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