NHS
Expanded Screening Pilot

Results of the Newborn Blood Spot Screening

Parent Information Sheet Maple Syrup Urine Disease (MSUD) - Confirmed Diagnosis

Your baby has been seen by the Specialist Metabolic Team and a diagnosis of MSUD has been confirmed. However, each child with MSUD is different and there may be some treatment differences from child to child.

This leaflet will help you to understand the condition and its treatment.

What is MSUD?

MSUD stands for Maple Syrup Urine Disease. MSUD is a rare disorder in which a baby or child has a problem breaking down protein. Protein is an essential part of our nutrition and is needed for growth and repair of tissues in the body. In order for the body to use protein from the food we eat, it is broken down into smaller parts called amino acids. Special enzymes (chemicals found naturally in your body) then make changes to the amino acids so that the body can use them.

People with MSUD are missing one of the enzymes that help break down some of the amino acids from the foods they eat and this causes harmful substances to build up in their blood and urine.

People with MSUD have problems breaking down three amino acids called leucine, isoleucine and valine from the food they eat and this causes harmful substances to build up in their blood and urine. High levels of these substances can cause an unusual sweet smell in the urine and sweat. MSUD can be treated to try and prevent the harmful build up of substances.

What are the symptoms of MSUD?

MSUD may affect different babies in different ways. Some babies start to develop symptoms in the first days or weeks of life. These are the symptoms of a developing metabolic crisis (a period of time in which the effects of the condition make your baby or child poorly) and include:

- Poor feeding
- Irritability
- Sleepiness
- Vomiting
- Breathing difficulties and fast breathing
- Coldness

Left untreated, babies with MSUD may deteriorate, have fits, slip into a coma, and risk dying. This is due to high levels of the harmful substances in the body. If levels are very high, intensive care and dialysis may be needed to bring the levels down as quickly as possible.

Sometimes a baby may not develop symptoms of a metabolic crisis until later on in the first year of life or even later during childhood. In this case, a crisis may be caused by having an infection such as an upset stomach or vomiting illness.

How is MSUD treated?

Treatment is given to children with MSUD to reduce the build up of toxins which can cause a metabolic crisis and learning difficulties. Dietary treatment and emergency treatment can be effective in preventing damage to the brain.

The special low protein diet is controlled by a Specialist Metabolic Dietitian. The aim of this diet is to reduce the amount of 3 amino acids called leucine, valine and isoleucine. These are found in foods such as meat, fish, cheese, eggs, pulses and nuts. They contain too much of these amino acids and therefore should be avoided. However, babies need some of these amino acids to grow and develop. Measured amounts of some protein containing foods (e.g. breast or baby milk, yoghurt & cereal).

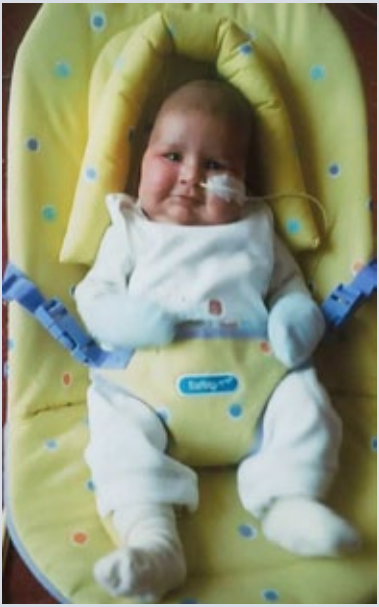
Foods that provide the body with protein include milk, meat, fish, cheese, eggs, pulses and nuts. All baby milks (including breast milk) contain more protein than can be tolerated by babies with MSUD. Breast feeding is still encouraged but the amount of milk a baby with MSUD is given will be measured and controlled. Your Specialist Metabolic Dietitian will advise you on this.

As high protein foods and milk have to be limited, a special baby milk which does not contain leucine, valine and isoleucine is given to meet requirements for protein, calories, vitamins and minerals. Your child may also require supplements of isoleucine and valine to maintain blood levels at the right level. Although isoleucine and valine cannot be broken down properly, if leucine is very restricted in the diet, it may cause the level of isoleucine and valine to fall too low. This supplement is very important because it allows normal growth and development, as well as helping to reduce the build up of harmful toxins to the brain. Your child will require regular blood tests to monitor the levels in their blood. Your Specialist Metabolic Dietitian will provide you with detailed information about special dietary products and will explain how much your baby should have. Your Specialist Metabolic Dietitian will also tell you about the availability of special low protein bread, biscuits, rice, flour and pasta.

What happens if my baby is ill?

Most babies and children will have infections with symptoms such as temperatures, coughs, colds, diarrhoea or vomiting from time to time. Babies and children with MSUD are at risk of having a metabolic crisis when they have infections, but the risk of having a crisis can be greatly reduced by starting a special feed called the emergency regimen. The Metabolic Dietitian will provide detailed instructions on how to give the emergency regimen. This involves stopping protein containing milk and food, replacing this with regular high sugar drinks (also known as glucose polymer powders i.e. SOS, Maxijul or Polycal). It is also very important to give the special amino acid formula, with valine and isoleucine supplements to help control the leucine levels in the blood.

What should I do if my baby vomits the emergency regimen drinks?



Many babies and children will be able to drink and keep down their emergency feeds. However sometimes a baby or child will vomit what they drink. If your baby continues to vomit or has repeated episodes of diarrhoea despite using the emergency feeds, you should take your baby to hospital immediately. It is helpful if you contact you Specialist Metabolic Team to say you are on your way, but do not delay going to hospital.

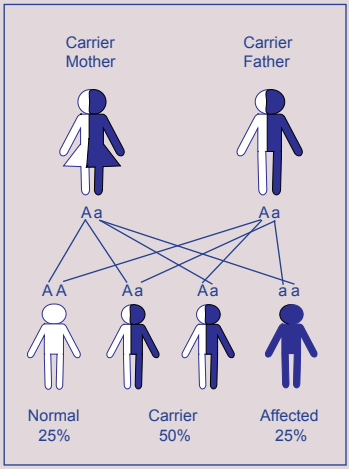
If you have to take your baby to hospital (Emergency Department (ED) or Accident & Emergency (A&E)), it is very important that you take your copy of the A&E guide to show the doctors as they may not be familiar with the treatment of MSUD. These emergency guidelines are also available on the BIMDG website (www.bimdg.org.uk).

If your baby does come to hospital with vomiting, their fluids can be given into their veins by a drip (intravenously) and they will be closely observed.

If your baby or child seems unusually sleepy, irritability or has rapid breathing, they should be taken to hospital immediately.

How will MSUD affect my child's future?

Before newborn screening, most cases of MSUD were identified after the baby or child had developed severe symptoms including a metabolic crisis or learning difficulties. Now that babies with MSUD can be detected earlier by newborn screening, specific treatment can be given earlier to improve the long term outlook.



Why does my baby have MSUD?

MSUD is an inherited condition. Everyone has two copies of the gene for MSUD. A baby with MSUD has inherited two faulty copies of the gene. The parents have one normal copy and one faulty copy and are said to be 'carriers'. When two MSUD carriers have a baby, they have a 1 in 4 (25 %) chance in each pregnancy of having a child with MSUD. There is nothing the parents could have done to prevent their child having MSUD. The diagram shows how this happens.

Will my other children need to be tested?

Your other children may be at risk of MSUD even though they may not have shown any symptoms to date. It is therefore very important that they are tested if they have not been previously screened for MSUD.

What about future children?

A new baby has a 1 in 4 (25%) risk of having MSUD and it is important to test at the earliest opportunity. You should inform the Midwife and Doctor looking after you that there is a history of MSUD in your family. You should ask for a referral to a paediatrician or genetic counsellor for advice then make a birth plan taking account of the information received (make sure the birth plan is written in your notes).

Who can I ask for advice and support?

The paediatric or metabolic clinician who is responsible for your child's care will be happy to discuss any queries you may have.

Contact Details for the Specialist Metabolic Team:

Specialist Centre		
Consultant		☎
Metabolic Dietitian		☎
Clinical Specialist Nurse		☎
Ward (if applicable)		☎

My notes:

Where can I find more information or support?

More information can be found at the Expanded Screening Programme website:

www.expandedscreening.org

CLIMB (The National Information Centre for Metabolic Diseases) provides information and support for people with MSUD and their families.

Climb Building
176 Nantwich Road
Crewe
CW2 6BG

Telephone helpline: 0800 652 3181 (Freephone) or 0845 241 2172

Website: www.climb.org.uk

Email: info@climb.org.uk

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