UK National Screening Committee

NHS Screening Programmes

Expansion of the Newborn Blood Spot Screening Programme – Frequently asked questions

What conditions has the UK National Screening Committee recommended including in the Newborn Blood Spot Screening Programme?

Currently all babies in the UK are offered screening for phenylketonuria (PKU), congenital hypothyroidism (CHT), sickle cell disease (SCD), cystic fibrosis (CF) and medium-chain acyl-CoA dehydrogenase deficiency (MCADD) through the NHS Newborn Blood Spot Screening Programme.

The UK National Screening Committee, reviewed the results of a study run by the National Institute of Health Research and Sheffield Children's NHS Foundation Trust. In this study, 430,000 babies across the country were screened for five rare diseases in addition to the current five for which every newborn is already screened.

Following a consultation, the UK National Screening Committee has recommended expanding the current newborn blood spot screening programme to include four new conditions; homocystinuria (HCU), maple syrup urine disease (MSUD), glutaric aciduria type 1 (GA1) and isovaleric acidaemia (IVA).

Babies affected by these inherited conditions have problems breaking down parts of proteins (amino acids) in the body. When the levels of these amino acids get very high, they can be harmful to the baby.

What are amino acids?

Amino acids are the building blocks of protein. Normally, people get protein from foods such as meats and pulses and the body uses them to help keep the tissues of the body healthy. Amino acids which are not needed are broken down and removed from the body.

What is screening?

Newborn blood spot screening aims to identify babies who are at high risk of having certain serious conditions before they develop symptoms. Screening is not the same as diagnosis; instead it identifies which babies need to go on to have further tests to determine whether or not they do have the condition. By detecting these conditions early it is possible to treat them and reduce their severity.

If HCU, MSUD, GA1 and IVA are not picked up early, they almost always cause severe developmental problems, including serious mental disability and possibly

coma, or even death.

How are babies identified as high risk of having these conditions?

Newborn blood spot screening is offered to all babies in the UK. The blood spot sample is taken on day 5, and in exceptional circumstances between day 5 and day 8 (day of birth is day 0). A health professional pricks the baby's heel and collects a small amount of blood onto the newborn blood spot card (a special filter paper, formerly known as the Guthrie card). The card is sent to the regional newborn screening laboratory and analysed for nine conditions (which will include these four new conditions).

What is homocystinuria (HCU)?

HCU is a rare disorder that prevents the breakdown of the amino acid, homocysteine.

Without treatment, most children with HCU have learning difficulties and problems with their eyes. They may also develop bone abnormalities (osteoporosis), blood clots, or strokes.

What is maple syrup urine disease (MSUD)?

A baby with MSUD has problems breaking down the amino acid known as leucine, isoleucine and valine.

Most babies with MSUD start to become unwell in the few days before they are born. They have problems such as poor feeding, vomiting and excessive sleepiness.

Without treatment, this can lead to a coma and permanent brain damage. In older children, a minor illness such as a chest infection or stomach upset can lead to serious problems.

What is glutaric aciduria type 1 (GA1)?

A baby with GA1 has problems breaking down the amino acids lysine and tryptophan.

In children with GA1, a minor illness, such as a chest infection or a tummy upset, can lead to serious problems and the need for immediate hospital treatment. Early signs may be vomiting, excessive sleepiness and breathing difficulties.

Without treatment the child can go into a coma and many patients die in early adulthood. If they recover from the coma there is a high likelihood of brain damage that can affect their ability to control muscles and movements. This means that they may be unable to sit, walk, talk or swallow.

What is isovaleric acidaemia (IVA)?

A baby with IVA has problems breaking down the amino acid leucine. For people with IVA, eating too much protein can cause harmful substances to build up in the blood.

Babies with IVA can become severely unwell. Early signs may be vomiting, excessive sleepiness, floppiness and rapid breathing.

Without treatment, IVA can lead to a coma and permanent brain damage. Some babies with IVA have problems within a few days of birth; other children become unwell at a few months or years of age, maybe during a minor illness, such as a chest infection or a tummy upset.

IVA can vary in severity – it is only severe cases that the screening programme aims to identify.

Who is at risk and how many are affected?

All four conditions are inherited conditions. This means that the diseases are transmitted in our genes and DNA. Where a baby has one of these rare conditions, it means that both the parents carry this gene.

These conditions are very rare:

- Homocystinuria (HCU) affects about 1 in every 150,000 births. Four or five babies with the problem are born every year in England
- Maple syrup urine disease (MSUD) affects just over 1 in every 100,000 births (that is seven babies a year born in England)
- Glutaric aciduria type 1 (GA1) affects about 1 in every 110,000 births
- Isovaleric acidaemia (IVA) affects about 1 in every 150,000 births

What treatment options are available for these conditions?

These conditions require lifelong management, through special diets and in some cases, supplementary vitamins or medicines.

Early treatment and careful management will allow affected babies to develop normally.

During illnesses, such as chest infections or stomach upsets, these children can become seriously unwell. Special drinks (called Emergency Regimen) are required at these times and hospital admission may also be needed.