P&N
Scottish Pregnancy and Newborn Screening Programme
NHS National Services Scotland
Spring 2017

# Expanded newborn bloodspot screening and the extension of the offer of screening for babies up to one year of age from 20 March 2017

## Four additional conditions to be included in the newborn bloodspot screening programme

**On the recommendation of the UK National Screening Committee (UK NSC) NHS Scotland shall be implementing screening for four additional Inherited Metabolic Disorders (IMD) as part of the newborn bloodspot screening programme from the 20th March 2017.**

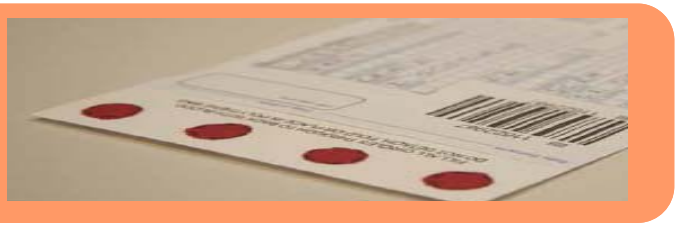
These are

- Maple Syrup Urine Disease (MSUD)
- Homocystinuria (HCU)
- Isovaleric acidaemia (IVA)
- Glutaric Aciduria type 1 (GA1)

Screening already takes place for 2 other IMD conditions, Phenylketonuria (PKU) and medium-chain acyl-CoA dehydrogenase deficiency (MCADD).

All of these conditions are a result of being unable to break down particular amino acids. Amino acids are the building blocks of protein and make up a large proportion of the human body. Normally, people get protein in foods such as meats 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. When the levels of these amino acids get very high, they are harmful.

The diseases have different effects, including seizures (fits), falling into a coma, brain damage and even death. Without treatment, babies can become suddenly and seriously ill. The most important aspect of treatment for these disorders is dietary and special diets have been designed for each condition Some may also need medication. During periods of common childhood illness, additional advice is given to parents and on rare occasions, admission to hospital may be needed.



Positive results are extremely rare for these disorders. They typically occur in between 1 in 100,000 to 1 in 200,000 births. As with all screening programmes, while most babies with these disorders will be detected, it is possible that a baby with one of the conditions may not give a positive result.

From the implementation of expanded screening on the 20th March 2017, the new patient information leaflet should be used and women should be informed that all six IMDs have to be screened for as a package and it will no longer be possible to screen for some of the IMDs and not the others; i.e. screening is for all 6 disorders or cannot take place. This is because the laboratory will be moving to an off the shelf pre-prepared analytical kit. Women can still make decisions on whether to be screened for sickle cell disease (SCD), cystic fibrosis (CF) and congenital hypothyroidism (CHT) on an individual basis.

It should be noted that it has not been possible to update the consent form within Scottish Woman Held Maternity Record (SWHMR) at this time therefore when obtaining written consent for the 6 metabolic conditions this should be hand written in the space left available. NHS Boards should make arrangements to update the electronic consent forms held within IT systems to remove the individual fields for PKU and MCADD and change this to screening for 6 Inherited Metabolic Disorders

## Key Dates

Pregnancy & Newborn Coordinators' meeting
6th March 2017
4th July 2017
9th November 2017

Pregnancy Screening Steering Group
22 Feb 2017

Newborn Blood Spot Screening Steering Group
28 Feb 2017

# Expanded Newborn Bloodspot Screening

Babies with **MSUD** are unable to break down the amino acids leucine, isoleucine and valine. Very high levels of these amino acids are harmful. One of the characteristic symptoms of MSUD is sweet-smelling urine, which gives the condition its name. Many babies with MSUD become unwell when they are a few days old, with:

- Poor feeding
- Vomiting
- Excessive sleepiness

Babies diagnosed with MSUD are first referred to a specialist metabolic dietician and given a low-protein diet. This is tailored to reduce the amount of amino acids the baby receives, especially leucine, valine and isoleucine.

Babies affected by **Homocystinuria (HCU)** cannot process the amino acid methionine, causing a build-up of methionine and a chemical called homocysteine. Without treatment, most children with HCU have learning difficulties and eye problems. They may also develop bones that are abnormally long and thin (osteoporosis), and blood clots or strokes.

In some babies, it is possible to control the levels of homocysteine with high doses of vitamin B6 (pyridoxine). Babies diagnosed with HCU that do not respond to vitamin B6 are referred to a specialist metabolic dietician and given a low-protein diet.

Babies affected by **Isovaleric Acidaemia (IVA)** cannot process the amino acid leucine. Early signs may be:

- Vomiting
- Excessive sleepiness
- Floppiness
- Rapid breathing

If IVA is diagnosed, treatment can be given straight away to reduce the risk of serious complications. Treatment includes a special diet, advice and sometimes medication.

IVA can vary in severity. In some mild forms of IVA, the risk of problems is much lower and this means that the treatment can be simpler.

Babies affected by **Glutaric Aciduria type 1 (GA1)** cannot break down the amino acids lysine, hydroxylysine and tryptophan. Normally, these amino acids are broken down into a substance called glutaric acid, which is then converted into energy. Babies with GA1 don't have the enzyme that breaks down glutaric acid, leading to a harmfully high level of this and other substances in the body. In children with GA1, a minor illness, such as a chest infection or a tummy upset, can lead to serious problems. Early signs may be:

- Vomiting
- Irritability
- Excessive sleepiness
- Floppiness
- Breathing difficulties

If a child with GA1 has these symptoms, they should be taken straight to hospital. Without treatment, the child can go into a coma. Though most children come out of the coma, they usually have brain damage that affects their ability to control their muscles and movements. This means that they may be unable to sit, walk, talk or swallow.

Babies diagnosed with GA1 are referred to a specialist metabolic dietician and given a low-protein diet. This is tailored to reduce the amount of amino acids the baby receives, especially lysine and tryptophan. They will also be prescribed a medication called L-carnitine, which helps to clear some of the excess glutaric acid.

## Important points

- All babies having the bloodspot sample taken on or after the 20th March should be offered expanded newborn screening
- Some new mothers may not have received the new booklet. To ensure informed choice, use the new booklet when obtaining consent.
- Screening is offered for all 6 IMDs as a package they cannot be screened for individually
- The other conditions can still be offered individually as normal
- Remember to use the new national patient information leaflet from the 20th March
- NHS Boards should update their IT fields for screening offer and outcomes and any electronically generated consent form to include the new conditions.
- Consent for screening for the 6 IMDs should be written in manually in SWHMR

# Important Service Change

## Extension of the offer of newborn bloodspot screening for babies up to one year of age.

Up until the 20 March the policy was to offer newborn bloodspot screening for those babies who did not have a documented result up until 6 months of age. **From the 20 March** screening should be offered for those babies up until 1 year of age. The policy applies equally to all infants resident in Scotland, whether born in Scotland other countries of the UK, or born abroad. This will bring NHS Scotland in line with the rest of the UK. The cut-off at a year of age is largely a pragmatic decision, backed up by professional consensus that there are still benefits of screening older infants and children, but these become less with age.

The screening test for CF is unreliable after a baby is 8 weeks old. Older babies with CF may have normal results and so this test will not be done in babies over 8 weeks of age. If a child has repeated chest infections or diarrhoea, medical advice should be sought and a test for cystic fibrosis may be arranged. This test is more complicated than the screening test and so it is not carried out on all babies.

If parents decide not to accept the offer of screening, or the baby is over twelve months old, this should be recorded in clinical notes and on the child health system. Parents should be asked to seek medical advice and remind staff that their baby has not been tested if their baby has signs of chronic health problems such as developmental delay, chronic diarrhoea or repeated lower respiratory tract infections.

If a child is under a year of age (up to but not including their first birthday) and has no documented results (or declines) for all five conditions screened for before the expansion of the programme, screening should be offered for all the untested conditions (including the four additional inherited metabolic diseases) **only** if the blood spot sample can be taken before they reach a year of age.

If for a child under a year of age, there are documented results (or declines) for all five conditions screened for in Scotland before the expansion of the programme (SCD, CF, CHT, PKU and MCADD), screening should **not** be offered for the four additional inherited metabolic disorders (MSUD, IVA, GA1 and HCU).

If parents accept screening and the blood spot sample is taken, the screening laboratory will perform all processes until screening has been completed for all the conditions – this includes processing initial samples received in the laboratory on or after the child's first birthday and requesting and processing repeat samples if required.

**NHS Boards should ensure that there are staff trained and responsible for taking the blood spots in infants that are no longer the responsibility of the midwifery unit.**

## National Screening Committee News

The annual evidence report summarising all the screening recommendations made by the NSC in 2015-16 can be found here :
https://www.gov.uk/government/publications/uk-national-screening-committee-recommendations-annual-report

**20 Years of the UK NSC**
The recent UK National Screening Committee (UK NSC) conference held in December 2016 celebrated 20 years of the UK NSC. A summary of the event and the links to the presentations on the day can be found here
https://phescreening.blog.gov.uk/2016/12/21/20-years-of-the-uk-nsc-thoughts-on-the-2016-annual-stakeholder-conference/

# Useful Resources

## NHS Scotland websites.

- **Health Professionals**

http://www.pnsd.scot.nhs.uk/

- **Public**

https://www.nhsinform.scot/healthy-living/screening/pregnancy-screening/introduction-to-pregnancy-screening

https://www.nhsinform.scot/healthy-living/screening/newborn-screening/introduction-to-newborn-screening

## NHS England websites.

- **Health Professionals**

The NHS England main site for screening programmes can be found here for:

https://www.gov.uk/topic/population-screening-programmes

The English screening programme centres are no longer producing newsletters. Instead you can sign up for the PHE screening blog alert service here

https://phescreening.blog.gov.uk/

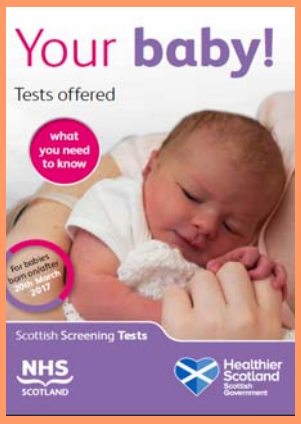
# Reviews

Scottish Down's Syndrome Laboratories - Recommendations endorsed by the Cabinet Secretary and implementation underway

# News

## New Patient Information leaflets

**For use from the 20th March 2016**


# Networks

## Networks to be involved in...

SPAIIN—Scottish Paediatric & Adolescence Infection & Immunology Network

http://www.spaiin.scot.nhs.uk/

Paediatric Cystic Fibrosis

http://www.knowledge.scot.nhs.uk/child-services/communities-of-practice/cystic-fibrosis.aspx

Scottish Managed Clinical Network for Inherited Metabolic Disorders in Scotland (IMD Scotland)

http://www.imd.scot.nhs.uk/

# Supporting resources...

- Updated service specification
- Updated protocols
- Updated laboratory handbook
- Updated bloodspot sampling guidance
- Updated Child Health Records guidance
- New patient information leaflets

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