Directorate of Chief Medical Officer and Public Health

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The Scottish Government
Riaghaltas na h-Alba

Dear Colleague

IMPORTANT CHANGES TO THE NEWBORN SCREENING PROGRAMME IN SCOTLAND

1. I am writing to inform you of the extension of the Newborn Blood Spot Screening programme to test for Homocystinuria (HCU), Maple Syrup Urine Disease (MSUD), Glutaric Aciduria type 1 (GA1), and Isovaleric Aciduria (IVA) from **20 March 2017,** following review by the UK National Screening Committee (UK NSC).

2. From this date, all newborn babies up to one year should be offered (age appropriate) screening (extended from the current 6 months).

Background

3. Newborn blood spot screening identifies babies who may have rare but serious conditions. Most babies screened will not have any of the conditions but, for the small number that do, the benefits of screening are enormous. Early treatment can improve their health and prevent severe disability or even death.

4. Currently all babies in Scotland are screened for Phenylketonuria (PKU), Congenital hypothyroidism (CHT), Sickle cell disease (SCD), cystic fibrosis (CF) and Medium-chain acyl-CoA dehydrogenase deficiency (MCADD).

5. Based on the evaluation of a one year project in England that involved over 400,000 babies, the UK NSC has recommended that every baby in the UK should be screened for the additional four conditions. Like PKU and MCADD, the additional conditions are autosomal recessive inherited metabolic diseases (IMDs).

From the Chief Medical Officer
Dr Catherine Calderwood MA MRCOG FRCP(Edin)

Date: 10 February 2017

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Addresses
For action
Chief Executives, NHS Boards
Directors of Public Health, NHS Boards

For information
Medical Directors, NHS Boards
Chairs, NHS Boards
Directors of Nursing & Midwifery, NHS Boards
General Practitioners
Consultant Paediatricians
Scottish Newborn Screening Laboratory
Primary Care Leads, NHS Boards
Pregnancy and Newborn Screening Co-ordinators
Practice Nurses
Health Visitors
Carol Colquhoun, National Services Division
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Further Enquiries

Policy Issues

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Implementation

6. The introduction of these changes to the Newborn Blood Spot Screening programme requires careful planning as it is introduced across Scotland on the same date. This has been subject to discussion and the following has been agreed by the Expanded Blood Spot Project Group:

- No additional blood spots are required. Four good quality blood spots are still required.
- From 20 March 2017 all NHS Boards must have in place systems/processes to ensure that appropriate consent is obtained for the full range of newborn screening tests and recorded on the consent form or IT systems, in line with local arrangements in place. NHS Boards should ensure that all relevant staff have been informed and aware of their roles and responsibilities for the implementation of the additional screens. NHS Boards must also ensure that referral pathways are agreed and in place.
- From 20 March 2017 the Scottish Newborn Screening Laboratory will report tests for Homocystinuria (HCU), Maple Syrup Urine Disease (MSUD), Glutaric Aciduria type 1 (GA1), and Isovaleric Aciduria (IVA)

7. Annex A includes a summary of additional important information for health professionals involved in newborn blood spot screening. Further details are available online at www.pnsd.scot.nhs.uk.

Conditions

8. The four additional conditions are very rare. They typically occur in between 1 in 100,000 and 1 in 200,000 births. In general, early dietary-based treatment for these conditions is effective. If untreated, babies with MSUD, IVA and GA1 can become suddenly and seriously ill, while symptoms of HCU can take up to one or two years to emerge. Serious effects, including severe mental retardation or death, may result if untreated.

Communications

Information for professionals

9. Information and guidance for professionals to support the developments will be published on the pregnancy and newborn website as it becomes available www.pnsd.scot.nhs.uk.

10. A newsletter will be available and distributed through the usual channels to advise of the new arrangements.

Public information materials

11. The updated public information leaflet aimed at parents to help support informed choice, will be available to NHS Boards by mid March 2017. The leaflet is designed to be used as a tool to support the health professional in a discussion about screening and the taking of informed consent from the parent.

12. The required quantities of the leaflets can be ordered from your local health promotion department. Any general questions in relation to distribution should be directed to the Publications Team at NHS Health Scotland on 0131 314 5300 nhs.healthscotland-publications@nhs.net.

13. The leaflet, translations, as well as an easy read version will be available to view/download from the Health Scotland website at: (http://www.healthscotland.com/topics/health/screening/pregnancynewborn.aspx.). NHS Health Scotland is happy to consider requests for other languages and formats. Please contact 0131 314 5300 or email nhs.healthscotland-alternativeformats@nhs.net.

14. Further information for the public about the pregnancy and newborn screening programmes can also be found online at:

www.nhsinform.scot/healthy-living/screening/newborn-screening/introduction-to-newborn-screening.
www.readysteadybaby.org.uk.

Education and training

15. Education and training for health professionals is being organised at local level through NHS Board screening co-ordinators and screening leads and supporting resources will be available at www.pnsd.scot.nhs.uk.

Yours sincerely

Dr Catherine Calderwood
Chief Medical Officer

Detailed information on the new conditions included in the bloodspot screening test

Homocystinuria

Homocystinuria causes a build-up of the amino acid homcysteine in blood and urine. Left untreated it can cause bone damage, visual problems and brain damage. These symptoms can usually be prevented with prompt diagnosis and treatment.

Maple syrup urine disease

Maple syrup urine disease is a rare disorder affecting around 1 in 185,000 children. It disrupts the normal functioning of amino acids inside the body. Symptoms can range from the relatively mild, such as vomiting, to severe, such as seizures and coma. The condition can normally be controlled through a specialist diet.

Glutaric aciduria type 1

Glutaric aciduria type 1 is a genetic condition associated with amino acids dysfunction. Symptoms include muscle spasms and bleeding inside the eyes and brain. The condition can be treated using a combination of medication and occupational therapy.

Isovaleric acidaemia

Isovaleric acidaemia is another genetic amino acid disorder. Initial symptoms include sweaty feet, but without treatment the condition can rapidly worsen, leading to seizures and, in some cases, coma. The condition can be successfully treated using a diet plan designed to avoid certain proteins.