From the Chief Medical Officer
Dr Michael McBride

Department of Health
www.health-ni.gov.uk

Castle Buildings
Stormont
BELFAST
BT4 3SQ
Tel: 028 9052 0563
Fax: 028 9052 0574
Email: michael.mcbride@health-ni.gov.uk

Your Ref:
Our Ref: HSS(MD) 13/2016
Date: 26 July 2016

HSS(MD) 13/2016

For Action:

Chief Executive PHA *(for onward cascade to all relevant Public Health Staff)*
Chief Executives HSC Trusts *(for onward cascade to all relevant staff)*
Chief Executive HSCB *(for onward distribution to GP Medical Advisers)*
All General Practitioners and GP Locums (*for onward distribution to relevant practice staff*)

Dear Colleague

EXPANSION OF THE NEWBORN BLOOD SPOT SCREENING PROGRAMME

ACTION REQUIRED

Public Health Agency should arrange for the establishment of a regional group to oversee the implementation of these additions to the Newborn Blood Spot Screening Programme. They should also identify a lead professional to co-ordinate implementation at Trust level.

Chief Executives of Health and Social Care Trusts should ensure that arrangements are in place:

- for MSUD, HCU, GA1 and IVA to be added to the existing Newborn Blood Spot Screening Programme from October 2017;
- introduce a limited Saturday service in the newborn screening laboratory for the review of screening results, with appropriate diagnostic follow up and clinical referral, from October 2017;
- for the Newborn Blood Spot Screening Programme to be delivered to the standards and protocols agreed by the UK National Screening Committee; and
- for quality assurance of the programme including ongoing monitoring and auditing.
- ensure that arrangements are in place for the management of babies with a positive diagnosis.

Summary

1. The purpose of this letter is to inform health professionals of four additional conditions that are to be added to the Newborn Blood Spot Screening Programme in Northern Ireland from October 2017. These

are maple syrup urine disease (MSUD), homocystinuria (HCU), glutaric aciduria type 1 (GA1), and isovaleric acidaemia (IVA).

2. These rare but potentially serious inherited condition mean the body can't process certain amino acids (the "building blocks" of protein), causing a harmful build-up of substances in the blood and urine. Normally, a person's body can break down protein foods such as meat and fish into amino acids. Any amino acids that aren't needed are usually broken down and removed from the body.

Background

3. The aim of the Newborn Blood Spot Screening Programme is to prevent disability and death, from a range of metabolic conditions, through early diagnosis and effective interventions. Screening involves putting a small heel-prick sample of a baby's blood onto a card which is then analysed in a laboratory.

4. The Newborn Blood Spot Screening Programme currently in place offers screening for phenylketonuria (PKU), congenital hypothyroidism (CH), cystic fibrosis (CF), MCADD (medium chain acyl CoA dehydrogenase deficiency), and sickle cell disorders (SCD).

5. The UK National Screening Committee (NSC) has recommended that every newborn baby in the UK should be screened for four additional conditions. This means expanding the existing Newborn Bloodspot Screening Programme to include screening for: Homocystinuria (HCU), Maple Syrup Urine Disease (MSUD), Glutaric Aciduria Type 1 (GA1) and Isovaleric Acidaemia (IVA). This recommendation is supported by the Northern Ireland Screening Committee. The Minister of the Department of Health has agreed with the recommendations and the additional funding required has now been identified.

Resources

6. A number of pieces or preparatory work will need to be undertaken before the additional conditions that are to be added to the Newborn Blood Spot Screening Programme can be fully implemented. These include changes to the IT and failsafe systems; development of new guidelines and standard operating procedures for clinical and laboratory staff, and revision of the pre-screening parents' information leaflet to include the additional conditions. The estimated cost of this preparatory work in 2016/17 is £70k and has been allocated to the Public Health Agency to allow this preparatory work to begin.

7. The recurrent revenue cost of the expansion of the programme is estimated to be approximately £100k per annum and this funding will be factored into planning assumptions from 2017/18.

Implementing the changes to the Blood Spot Screening Programme

8. All Health and Social Care Trusts should begin make the necessary arrangements to add screening for MSUD, HCU, GA1 and IVA to the existing Newborn Blood Spot Screening Programme from October 2017**.**

Maple Syrup Urine Disease (MSUD)
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.

Homocystinuria (HCU)
Babies with HCU are unable to fully break down the amino acid homocysteine. This can lead to harmful substances building up in the blood.

Glutaric Aciduria Type 1 (GA1)
Babies with GA1 are unable to break down the amino acids lysine and tryptophan. For people with GA1, eating protein can cause harmful substances to build up in the blood and urine.

Isovaleric Acidaemia (IVA)
Babies with IVA are unable to fully break down the amino acid leucine. For people with IVA, eating protein can cause harmful substances to build up in the blood.

Action

9. The Public Health Agency should arrange for the establishment of a regional group to oversee the implementation of these additions to the Newborn Blood Spot Screening Programme. They should also identify a lead professional to co-ordinate implementation at Trust level.

10. Chief Executives of Health and Social Care Trusts should ensure that arrangements are in place:
    - for MSUD, HCU, GA1 and IVA to be added to the existing Newborn Blood Spot Screening Programme from October 2017;
    - introduce a limited Saturday service in the newborn screening laboratory for the review of screening results, with appropriate diagnostic follow up and clinical referral, from October 2017;
    - for the Newborn Blood Spot Screening Programme to be delivered to the standards and protocols agreed by the UK National Screening Committee; and
    - for quality assurance of the programme including ongoing monitoring and auditing.

Follow up of children with positive results

11. Most babies who are screened will not have any of these conditions but, for the small numbers who do, the benefits of screening are enormous. Early treatment can improve their health and prevent severe disability or even death.

12. Health and Social Care Trusts should ensure that arrangements are in place for the management of babies with a positive diagnosis. As the numbers of positive cases will be small this would be best delivered at a regional level.

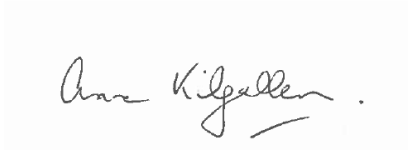
Quality Assurance and Monitoring

13. The Newborn Blood Spot Screening Programme should be commissioned and delivered in line with the protocols and standards which have been agreed by the UK National Screening Committee It should be subject to ongoing local performance management and audit and regional co-ordination of quality assurance. A minimum core of information should be collected on all infants to support performance monitoring of the programme, meeting of National standards and quality assurance. Appropriate failsafe mechanisms should be in place to ensure that screening is offered to all infants and those that 'screen positive' are followed up as appropriate.

Further information

14. For further information please contact Karen Simpson at karen.simpson@health-ni.gov.uk

Yours sincerely



pp DR MICHAEL McBRIDE
Chief Medical officer

CHARLOTTE McARDLE
Chief Nursing Officer

This letter is available on the DoH website at

www.health-ni.gov.uk/topics/professional-medical-and-environmental-health-advice/hssmd-letters-and-urgent-communications

CIRCULATION LIST

Director of Public Health/Executive Medical Director, Public Health Agency
Assistant Director Public Health (Screening), Public Health Agency
Director of Nursing, Public Health Agency
Medical Directors, HSC Trusts
Directors of Nursing, HSC Trusts
Director of Integrated Care, HSCB
Professor Patrick Johnston**,** Head of School of Medicine, Dentistry and Biomedical Science, QUB
Mr Keith Gardiner, Post Graduate Dean Northern Ireland Medical and Dental Training Agency
Dr Claire Loughrey, Director of Postgraduate General Practice Education, NIMDTA
Professor Hugh McKenna, Dean of Life and Health Sciences, University of Ulster
Professor Linda Johnston, Head of School of Nursing and Midwifery, QUB
Dr Owen Barr**,** Head of School of Nursing, University of Ulster (UU)
RQIA (*for onward transmission to all independent providers including independent hospitals*)