From the Chief Medical Officer
Dr Michael McBride

Department of
Health, Social Services and Public Safety
www.dhsspsni.gov.uk

AN ROINN
Sláinte, Seirbhísí Sóisialta agus Sábháilteachta Poiblí

MÄNNYSTRIE O
Poustie, Resydènter Heisin an Fowk Siccar

Chief Executives, HSS Boards
Chief Executives, HSC Trusts – *for onward distribution to lead directors for newborn bloodspot screening, laboratory services, genetic services and child health services*
Directors of Public Health, HSS Boards – *for onward distribution to newborn bloodspot screening leads*
Medical Directors, HSC Trusts – *for onward distribution to consultants in paediatrics, clinical chemistry, haematology and genetics*
Directors of Nursing, HSS Boards
Directors of Nursing, HSC Trusts – *for onward distribution to midwives and health visitors*
All GP units – *for onward distribution to all general practitioners*

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HSS(MD) 30/2008

Dear Colleague

ADDITIONS TO THE NEWBORN BLOODSPOT SCREENING PROGRAMME

Summary

1. The newborn bloodspot screening programme currently in place across Northern Ireland offers screening for phenylketonuria (PKU), congenital hypothyroidism (CH), cystic fibrosis (biochemical screening only), tyrosinaemia and homocystinuria. The UK National Screening Committee has recommended that screening for Medium Chain Acetyl CoA Dehydrogenase Deficiency (MCADD) and neonatal sickle cell disorders are added to the bloodspot programme, and that genetic testing is added to the cystic fibrosis (CF) screening protocol. Health and Social Services Boards and Health and Social Care Trusts should make arrangements for the implementation of MCADD screening from April 2009 and sickle cell screening from April 2010. Changes to the CF protocol should be in place from April 2009.

Background

Medium Chain Acetyl CoA Dehydrogenase Deficiency (MCADD)

2. MCADD is a rare inherited metabolic disorder which affects around one in 10,000 babies. The abnormality leads to an inability to metabolise sufficient energy from fat during periods of metabolic stress. It is a recognised cause of unexpected death in infancy and of acute encephalopathy in infancy requiring intensive care. If the disease is not identified and treated at an early stage around a quarter of affected children will

die from the condition, with one third of survivors sustaining significant permanent neurological damage. Early identification of the condition allows for the introduction of appropriate dietary management and feeding regimes.

Sickle Cell Disorders

3. Sickle cell disorders are inherited disorders which affect the red blood cells. If a baby has a sickle cell disorder the normally round red blood cells can change to a sickle or crescent moon shape under certain situations of stress. This will result in pain and can also cause severe anaemia, susceptibility to infection, damage to major organs and in some cases death. Early identification means that babies with sickle cell disorders can receive early treatment, including immunisations and antibiotics, which, along with parent education, will help prevent serious illness and allow the child to live a healthier life.

4. The incidence of sickle cell disorders in the UK has increased greatly. The conditions is more common people of African-Caribbean origin and from sub-Saharan regions but is also found in those with Arab, Mediterranean and Indian origins, as well as other groups in the UK.

Cystic Fibrosis (CF)

5. Cystic Fibrosis is an inherited disorder which affects about 1 in 2,500 babies. It can affect digestion and the lungs. Babies with the condition may not gain weight and have frequent chest infections. Although a child with CF may still become very ill, early treatment may help them live longer, healthier lives. For over 20 years newborn babies in Northern Ireland have been offered CF screening, using a biochemical test. The National Screening Committee has recommended a CF screening programme based on biochemical testing followed by genetic testing. Genetic testing will however identify a small number of babies who are CF carriers.

UK Newborn Screening Programme Centre

6. The UK Newborn Screening Programme Centre was established in 2002 with the overall objective of assuring high quality screening services for babies and their parents, through the development of a quality assurance and performance management framework for the bloodspot screening programme. It has produced a number of standards which underpin the QA programme.

7. The Programme Centre, which is funded by the Department of Health in England on behalf of all four UK countries, is a collaboration between Great Ormond Street Hospital for Children NHS Trust, the Institute of Child Health and the Institute of Education. It co-ordinates a UK wide quality assurance programme in partnership with health professionals and parents and monitors and facilitates improvements in the quality of screening processes and their outcomes.

Current Arrangements in Northern Ireland for Bloodspot Screening

8. For many years the Health and Social Services Boards and Health and Social Care Trusts have had arrangements in place for the commissioning and provision of a quality assured bloodspot screening programme for phenylketonuria (PKU), congenital hypothyroidism (CH), cystic fibrosis (biochemical screening only), tyrosinaemia and homocystinuria.

Implementing the changes to the Bloodspot Screening Programme

9. All Health and Social Services Boards and Health and Social Care Trusts should make the necessary arrangements to add screening for MCADD to the existing bloodspot screening programme from April 2009 and sickle cell disorders from April 2010. Genetic testing should be included in the cystic fibrosis screening protocol from April 2009. All bloodspot screening programmes including MCADD, CF and sickle cell disorders should be provided to the standards set by the Programme Centre.

Resources

10. Revenue funding of £150,000 from 2008/09 and £250,000 from 2009/10 has been made available by the DHSSPS for the implementation and delivery of these additions to the existing newborn bloodspot screening programme and to support continuous quality management and quality assurance of the programme.

Action

11. Chief Executives of Health and Social Services Boards should arrange for the establishment of a regional group to oversee the implementation of these additions to the newborn bloodspot screening programme. They should also identify a lead professional to co-ordinate implementation at Board level. Health and Social Services Boards should ensure that arrangements are in place:

- for MCADD to be added to the bloodspot screening programme from April 2009;
- for genetic testing to be added to the cystic fibrosis screening protocol from April 2009;
- for sickle cell disorders to be added to the bloodspot screening programme from April 2010.
- for the bloodspot screening programme to be delivered to the standards and protocols agreed by the UK Newborn Screening Programme Centre.
- for quality assurance of the programme including ongoing monitoring and auditing.

12. Chief Executives of Health and Social Care Trusts should identify a health professional to lead the implementation of these additions at Trust level. Health and Social Services Trusts should ensure that arrangements are in place:

- for MCADD to be added to the bloodspot screening programme from April 2009;
- for genetic testing to be added to the cystic fibrosis screening protocol from April 2009;
- for sickle cell disorders to be added to the bloodspot screening programme from April 2010;
- for the bloodspot screening programme to be delivered to the standards and protocols agreed by the UK Newborn Screening Programme Centre;
- for co-ordinating the management and delivery of the bloodspot screening programme;
- for quality assurance of the programme including ongoing monitoring and auditing.

Follow up of children with positive results

13. Health and Social Services Boards and 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. A small number of babies who are carriers of the CF gene or the sickle cell gene will be identified through screening. Arrangements should be in place to ensure that parents are informed and counselled about the implications of this.

Quality Assurance and Monitoring

14. The bloodspot screening programme should be commissioned and delivered in line with the protocols and standards which have been agreed by the UK Newborn Screening Programme Centre. 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
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Yours sincerely

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