Implementation of expanded newborn bloodspot screening and offer of screen extended to up to one year of age

Policy changes commencing from the 20th March 2017

Currently the Scottish Newborn Blood Spot Screening Programme screened for 5 conditions – sickle cell disease (SCD), cystic fibrosis (CF), congenital hypothyroidism (CHT), and 2 Inherited metabolic Disorders - phenylketonuria (PKU) and medium-chain acyl-CoA dehydrogenase deficiency (MCADD).

From the **20th March 2017**, the programme is being expanded to come in line with England and Wales to screen for 4 more rare but serious inherited metabolic disorders (IMDs) – maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (HCU) – helping to prevent severe disability or even death for around 30 more babies in the UK every year.

From this date parents can decline screening for CHT, SCD, CF or for all 6 IMDs however it will no longer be possible to screen for the IMD conditions separately as the laboratory will be moving to a new assay kit.

For most, this means a midwife carries out the test in the home when the baby is 96-120 hours (4-5 completed days) old. In exceptional circumstances samples can be taken between day 5 and day 8 (day of birth is day 0).

In addition from the 20th March 2017 if babies miss out on screening at 4-5 days then they can still be screened up to 12 months old for 8 of the 9 conditions (the test for CF is only reliable up to 8 weeks of age).—previously this was only offered up to 6 months of age. This can include babies who were born abroad and those whose mothers declined screening immediately after birth. If verifiable documented results cannot be found, then screening should be offered – **provided the blood spot sample can be taken before their first birthday**.

If parents decide not to accept the offer of screening, or their baby is over a year old, this should be recorded. Parents of babies who have not been tested should be advised to remind staff if their baby then shows signs of chronic health problems such as developmental delay, chronic diarrhoea or repeated lower respiratory tract infections.

What do health professionals need to do?

- 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.
- From 20th March the new patient information leaflets should be used and the old leaflets returned for recycling.
- The blood spot sample should ideally be taken between 96-120 hours of life (4-5 completed days), day of birth is day 0, and sent on the day of sampling. In exceptional circumstances samples can be sent between day 5 and day 8).
- If the sample is taken on day 4 then how many completed hours old the baby was at the time of the sample being taken should be recorded in the comments section of the card.
- Parents may decline any or all of the tests (the 6 IMD conditions count as one test) and must be informed that their baby remains eligible for screening within the Programme up to the age of 1 year.
- 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

- A newborn blood spot card must be completed for all babies, even if all tests are declined.
- Every child up to one year of age who moves into an NHS Board and/or where no previous screening has been recorded is eligible for and should be offered relevant tests.
- 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 may be easier to obtain a venous sample if the child is older however this must be drawn into a plain syringe with no additives and then the blood should be applied directly to the card.
- Suspected cases will be referred to the Inherited Metabolic Disorders team who will coordinate the ongoing investigations and management of the baby with the local paediatric team

Other Considerations

- Good quality blood spot samples are vital to ensure babies with rare but serious conditions are identified and treated early. They prevent the need for avoidable repeat samples, which can cause anxiety for parents, distress to babies, delays in the screening process and a waste of NHS money.
- Public Health England have developed a free online learning module to support sample takers in taking good quality blood spots. The resource will enable practitioners to understand why blood spot quality matters and why repeat samples are requested. It is open to practitioners in Scotland and registration for the module can be found here https://cpdscreening.phe.org.uk/bloodspot-elearning please note that whilst the principles of obtaining the bloodspot and the descriptions of the conditions are the same there are differences between the programmes for example the cards used in Scotland are slightly different . Please refer to the Scottish documents for guidance on completing the bloodspot card and for the screening and referral pathways.

More information on the screening programmes can be found on the website for Health professionals http://www.pnsd.scot.nhs.uk/

Supporting Resources

- Updated service specification, protocols, bloodspot taking guidance and Child Health Standard Operating Procedures for Blood Spot Reporting
- Updated national patient information leaflet
- Updated website information
- Updated Screening pathways
- A resource for Health professionals providing information on the screening programmes