P&N

Scottish Pregnancy and Newborn Screening Programme

NHS National Services Scotland

Spring 2016

Second Edition

Welcome to the second edition of the Pregnancy and Newborn Screening Programme newsletter! In order to keep the service aware of local and national changes, good news and updates, please share this information with your colleagues. We welcome any feedback, and/or article submissions.

Cessation of screening for rubella susceptibility in pregnancy

Screening for rubella susceptibility in Scotland to stop from the 1st June 2016 following approval from Ministers

The UK National Screening Committee (UK NSC) reviewed the evidence for rubella susceptibility screening in 2003 and 2012 and on both occasions advised that screening for rubella susceptibility did not meet the UK NSC criteria for a national screening programme and should be discontinued for the following reasons:

- The extremely low incidence of congenital rubella syndrome does not indicate the need for a population screening programme and is at a level defined as eliminated by the World Health Organisation.
- The test may falsely reassure some women that they are not susceptible to rubella infection during that pregnancy
- The focus on antenatal screening and postnatal immunisation may not be the optimum intervention to address rubella susceptibility as a public health issue in the wider population
- Alternative approaches to high levels of immunity in the total population may be more effective to ensure that women are protected against rubella infection prior to pregnancy

The UK NSC and the JCVI further reported that stopping antenatal screening is extremely unlikely to increase congenital rubella syndrome rates and that the population level of uptake of the first dose of MMR is now sufficient to stop screening.

Evidence

The evidence based reasons to end screening for rubella susceptibility include:

- Stopping antenatal screening is unlikely to increase rates of rubella. There are very few cases of rubella in the UK – with 12 cases of congenital rubella occurring in the decade 2005-2015. None of which could have been prevented by the screening programme
- Screening for rubella susceptibility in pregnancy does not give any protection to the unborn baby
- Being fully immunised with the MMR vaccine before becoming pregnant is the most effective way of ensuring women are protected against rubella in pregnancy

Policy Decision

Ministers have agreed that rubella susceptibility screening in pregnancy should cease in Scotland from 1 June 2016. The decision to end screening for rubella susceptibility in pregnancy has been made possible by the high levels of vaccination rates achieved through the MMR vaccination programme. Figures in Scotland show that 95% of children aged up to two years had received at least one MMR vaccination; with 97% having received at least one dose of MMR by five years.

Update

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/

Key Dates

Pregnancy & Newborn Coordinators' meeting
9th March 2016
24th August 2016
2nd November 2016

Pregnancy Screening Steering Group
14 March 2016

Newborn Blood Spot Group
18 April 2016

Important Service Change

Rubella Susceptibility in pregnancy screening

Rubella is a viral infection that causes a rash and fever. Infection during pregnancy can lead to serious health problems for the unborn baby. However, the success of the measles, mumps and rubella (MMR) vaccination means the disease is now incredibly rare.

The key points for maternity services to note are:

- the offer of antenatal screening should stop for all women in Scotland **booking on or after 1 June 2016**
- screening for HIV, hepatitis B and syphilis should continue to be offered to all eligible women
- the rubella screening pathway should be completed for women booking and accepting screening before 1 June, meaning:
- laboratories will test decreasing numbers of samples in July, August and September 2016 and will need to adjust assay supplies accordingly
- continued reporting and follow-up of results for women booking into this cohort by screening teams
- continued administration of postnatal MMR by maternity services and referral to primary care for second vaccination for non-immune women in this booking cohort

We are updating our protocols and operational handbooks. They will be published shortly. We are currently updating other resources, including patient information leaflets and website content, working collaboratively with our information and education teams.

Cell Free DNA testing (cfDNA) or Non Invasive Prenatal Testing (NIPT)

The UK NSC has recommended that Non-invasive prenatal testing (now more commonly called cfDNA, or cell-free DNA testing) should be introduced to the existing fetal anomaly screening programme (FASP) as a second stage screen for trisomies T21/T18/T13 (Down's, Edwards' and Patau's syndromes), following a risk score of 1 in 150 or greater in the combined test.

While the Scottish Government are fully committed to ensuring that pregnant women in Scotland have access to high quality, safe and effective screening services, there are a number of questions about the use of a Non-Invasive Prenatal Test (NIPT) that require further examination. No change will be made to the pregnancy screening programme without full consideration of the clinical evidence.

Expanded Newborn Bloodspot screening

The UK National Screening Committee (UK NSC) has recommended that every baby in the UK should be offered screening for an additional four inherited metabolic diseases (IMDs) as part of newborn blood spot screening. These are:

- maple syrup urine disease (MSUD)
- homocystinuria (HCU)
- isovaleric acidaemia (IVA)
- glutaric aciduria type 1 (GA1)

These are rare but serious inherited conditions

Babies affected by **Maple syrup urine disease (MSUD)** cannot process certain amino acids (the "building blocks" of protein), causing a harmful build-up of substances in the blood and urine. 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. 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. This can be harmful. 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 (. 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.

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. 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.

NHS Scotland supports the recommendation to implement screening for these additional conditions and work is ongoing in preparation for this. Communications on the timelines for implementation will be sent out as soon as possible.

National Screening Committee News

A new report *Screening in the UK: making effective recommendations* has been published highlighting the work during 2014-15 of the UK National Screening Committee (UK NSC).

https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/480961/UK_NSC_evidence_report_201415_online_version.pdf

This 2015 report summarises the UK NSC's work during a year when it made recommendations on 16 topics and acted on the House of Commons Science and Technology Committee's recommendations on screening.

Useful Resources

NHS Scotland websites.

- **Health Professionals**
http://www.pnsd.scot.nhs.uk/

- **Public**
http://www.nhsinform.co.uk/screening/overview/leaflets/#pregnancy

http://www.nhsinform.co.uk/screening/overview/leaflets/#pregnancy

NHS England websites.

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

- Health Professionals
https://www.gov.uk/topic/population-screening-programmes
- Public (in England):
http://www.nhs.uk/Livewell/Screening/Pages/screening.aspx

Reviews

Scottish Down's Syndrome Laboratories - Awaiting Board Chief Executives consideration

Evidence News

In current consultation:

- Kernicterus

http://legacy.screening.nhs.uk/kernicterus

- Scoliosis

http://legacy.screening.nhs.uk/scoliosis

- Toxoplasmosis

http://legacy.screening.nhs.uk/toxoplasmosis

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

Upcoming sites:
IMD
Congenital cardiac network

Get Involved...

If you have a suggestion for a topic for the next Newsletter and/or would be willing to submit a brief article, please email roxannegallacher.nhs.net

Please share this Newsletter with others you think may be interested. Thank you.

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