Chief Medical Officer Directorate

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Dear Colleague

IMPORTANT CHANGES TO THE SCOTTISH PREGNANCY SCREENING PROGRAMME

This letter sets out a number of changes and developments to strengthen and extend the Scottish pregnancy screening programmes. These changes take account of the updated advice from the UK National Screening Committee. The developments will be implemented from 28 September 2020.

In summary, these changes are:

- The expansion of the first trimester screening programme for Down's syndrome (Trisomy 21) to include screening for Edwards' syndrome (Trisomy 18) and Patau's syndrome (Trisomy 13).

- An evaluative roll out of Non Invasive Prenatal Testing (NIPT) as a second line screening test for those pregnancies with a higher chance result from a primary screen in the first or second trimester.

- The expansion of the screening programme for twin pregnancies to include combined first trimester screening for Down's syndrome, Edwards' syndrome and Patau's syndrome and second trimester quadruple screening for Down's syndrome and NIPT as a second line screen for those pregnancies receiving a higher chance result from the primary screen.

1. Action

NHS Boards will be responsible for implementing the changes in maternity services including the staff training required to deliver these improvements. Support from the National Services Division (NSD) will be available.

The Boards will also be responsible for the local performance management of the programmes and submitting data to Public Health Scotland against the Key Performance

From the Interim Chief Medical Officer
Dr Gregor Smith

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

For information
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NHS Boards, Maternity Clinical Directors
NHS Boards, Heads of Midwifery
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Indicators for national reporting against the Pregnancy and Newborn Screening Programmes.

Public Health Scotland will develop and produce the required patient information material.

They will also analyse and report on national data for the Scottish pregnancy and newborn screening programmes.

NHS Education Scotland has developed training materials for health professionals involved in the pregnancy and newborn screening programmes and providing care to pregnant women.

Healthcare Improvement Scotland will revise and review the clinical standards for pregnancy and newborn screening.

2. Background

At present, all pregnant women in Scotland are offered screening for Down's syndrome either in the first or second trimester. In the first trimester of pregnancy, the screen includes the measurement of biochemical markers in the mother's blood combined with the ultrasound measurement of nuchal translucency in the fetus. The second trimester screening test includes the measurement of 4 biochemical markers. If results indicate that the pregnancy is at a higher (more than a 1 in 150) chance of having a baby with Down's syndrome women are then offered follow-up diagnostic tests, either amniocentesis or chorionic villus sampling.

Edwards' Syndrome (Trisomy 18) and Patau's syndrome (Trisomy 13) are rare but very serious genetic conditions which affect a small number of babies every year. Although Edward's Syndrome and Patau's Syndrome may be identified through the current second trimester fetal anomaly scan, first trimester screening helps provide an earlier diagnosis allowing the woman to make informed decisions about her pregnancy at an earlier stage. See **Annex A** for more information on these conditions.

NIPT as an additional screen could reduce the number of women undergoing invasive diagnostic testing, which carries a risk of miscarriage. NIPT identifies DNA from the placenta in the mother's blood to assess the chance of the baby having Down's, Edwards' or Patau's syndrome.

Recognising that the use of such tests is relatively new, the UK NSC recommended NIPT should be offered as an additional test – to provide choice for pregnant women – with ongoing evaluation. This evaluative rollout will give a better understanding of the impact it will have on the decisions women and their partners make following positive test results.

The original evidence review on NIPT by the University of Warwick was re-considered by the UK NSC in January 2020 and they indicated that NIPT could also be offered in twin pregnancies as part of the evaluative roll out.

3. Implementation

The introduction of NIPT for Down's syndrome, Edwards' syndrome and Patau's syndrome to the Pregnancy Screening Programme requires careful planning as the test is scheduled to be introduced across Scotland on the same date. The Pregnancy and Newborn Screening

Programme Board has been working closely with NHS Boards and Public Health Scotland to ensure that:

- From 28 September 2020 all NHS Boards have in place systems and processes to ensure that appropriate consent is obtained for the full range of pregnancy screening tests and recorded on consent forms or IT systems, in line with local arrangements in place.

- NHS Boards should ensure that all relevant staff have been informed and completed the education package and are 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 28 September 2020 women booking in the first trimester with singleton or twin pregnancies can choose to have:
  - No screening
  - Screening for T21, T18 and T13
  - Screening for T21 only
  - Screening for T13/18 only

- Second trimester screening will continue to be for Down's syndrome but will now be offered for both singleton and twin pregnancies.

- NIPT is offered to pregnant women in accordance with recommendations for all three conditions as a second line screening option.

- Monitoring and evaluation data will be collected by each NHS Board and screening laboratory and submitted to Public Health Scotland.

Annex A includes additional information for healthcare professionals on Edwards' syndrome and Patau's syndrome, as these will be introduced into the current pregnancy screening programme at the same time as NIPT.

4. Communications

Information and guidance for professionals to support the developments will be distributed by National Services Division and published on NHS Inform pages.

5. Public Information

The updated public information leaflets aimed at parents to help support informed choice will be available to NHS Boards as a PDF by the end of August 2020 and as a hard copy leaflet by mid-September 2020. 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 parents.

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

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

Further information for the public about the pregnancy and newborn screening programmes can also be found online at:
www.healthscotland.scot/pschanges
www.nhsinform.scot/pregnancyscreening
www.nhsinform.scot/newbornscreening

6. Education and Training

Education and training for health professionals is being organised at national level by NHS Education for Scotland and supporting resources are available through TURAS.
https://learn.nes.nhs.scot/33524/women-children-and-famillies/expanded-pregnancy-screening-pathways-and-non-invasive-prenatal-testing-nipt

NHS Boards are asked to bring this letter to the attention of all those who will be involved in the implementation of the new screening test.

Yours sincerely

Dr Gregor Smith

Dr Gregor Smith
Interim Chief Medical Officer

Detailed information on the new conditions included in the pregnancy screening programme

Edwards' syndrome (Trisomy 18)

Edwards' syndrome, also known as Trisomy 18, is a rare but serious genetic condition caused by an additional copy of chromosome 18 in some or all of the cells in the body. There are different forms of Edwards' syndrome known as full, partial, and mosaic forms. Some of these forms may be more serious and screening cannot always tell how serious a baby's condition will be before they are born.

Babies with Edwards' syndrome can have a wide range of health issues, some of which are serious. These include:

- heart issues
- Difficulty swallowing and feeding
- breathing issues that require medical help
- hearing loss
- low birth weight and slow postnatal growth
- hernias in the wall of their stomach (where internal tissues push through a weakness in the muscle wall)
- bone anomalies – such as a curved spine
- frequent infections of the lungs and urinary system
- Most will have severe learning disabilities

Sadly, most babies with Edwards' syndrome do not survive, with 7 in 10 pregnancies diagnosed at 12 weeks of pregnancy ending in miscarriage or stillbirth. Some babies with less severe types of Edwards' syndrome, such as mosaic or partial trisomy 18, do survive with around 5 in 10 (52.5%) living longer than 1 week and around 1 in 10 (12.3%) living longer than 5 years. Babies with partial or mosaic forms may live into adulthood but will require ongoing care and support.

Patau's syndrome (Trisomy 13)

Patau's syndrome, also known as Trisomy 13, is a rare but serious genetic condition caused by an additional copy of chromosome 13 in some or all of the cells of the body. There are different forms of Patau's syndrome known as full, partial, or mosaic forms. Some of these forms may be more serious and screening cannot always tell how serious a baby's condition will be before they are born.
Babies with Patau's syndrome can have a wide range of health problems which are noted below:

- Low birth weight due to restricted growth in the womb
- Severe heart defects
- Holoprosencephaly (where the brain does not divide into 2 halves)
- Ocular anomalies such as small or absent eyes
- Cleft lip and palate
- Kidney issues and frequent urinary tract infections
- Difficulty swallowing, feeding, and breathing
- Seizures

- Omphalocele (where intestines, liver, or other organs are formed outside the body in a thin transparent sac)
- Moderate to severe learning disabilities and delayed physical development

Around 7 in 10 pregnancies diagnosed with Patau's syndrome at 12 weeks will end in miscarriage or still birth. Of all babies born with Patau's syndrome around 4 in 10 (43.1%) will live longer than 1 week and 1 in 10 (9.7%) will live longer than 5 years. Babies with partial or mosaic forms may live into adulthood but will require ongoing care and support.