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Dear Colleague

CHANGES TO THE DOWN'S SYNDROME SCREENING PROGRAMME

This letter sets a number of developments to strengthen the Down's syndrome screening programme. These changes take account of the updated advice from UK National Screening Committee (NSC). Further information on the changes can be found at annex A.

The main changes required to ensure Scotland aligns with NSC protocol are summarised as follows:

- To meet national performance standards, report of 'high chance' of Down's syndrome at term will be issued at 1 in 150 or greater and this cut off will be adopted for both first and second trimester Down's syndrome screening.
- The gestation range for 2nd trimester Down's syndrome screening will change to 14 weeks + 2 days to 20 weeks + 0 days of pregnancy.

The changes will be implemented in Scotland on **26 September 2011**.

It should be noted that a UK National Screening Committee expert group will be reviewing whether first trimester combined screening for Trisomy 13 (Patau's syndrome) and Trisomy 18 (Edwards' syndrome) should be added to the current screening programmes.

NHS Boards will be responsible for implementing any changes in maternity health services required to accommodate the developments in the Down's syndrome screening policy.

NHS National Services Scotland, National Services Division, will lead, co-ordinate and support NHS Boards and the Scottish Down's syndrome screening laboratories in making the changes.

Healthcare Improvement Scotland will revise and review the clinical standards for Down's syndrome screening.

NHS Health Scotland will review the required patient information material and health professionals training materials and make any required amendments.

**From the Chief Medical Officer
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Addresses

For action

Chief Executives, NHS Boards
Directors of Public Health, NHS
Boards

For information

Medical Directors, NHS Boards (to
cascade to Practice Managers)
Nurse Directors, NHS Boards
Chairs, NHS Boards
Royal College of General
Practitioners
Royal College of Nursing, Scottish
Division
Royal College of Midwives
Heads of Midwifery
General Medical Council
BMA

Further Enquiries

NHS Education Scotland to work with National Services Division to co-ordinate and lead a training update for health professionals involved in the Down's syndrome screening programme.

Yours sincerely

Harry Burns

HARRY BURNS

ANNEX A

Pregnancy Screening for Down's syndrome

Background

All pregnant women in Scotland are currently offered either a screening test in the first trimester of pregnancy in which measurement of biochemical markers in the mother's blood is combined with the ultrasound measurement of nuchal translucency in the fetus or, for those women who do not present early enough in their pregnancy to take advantage of first trimester screening, a second trimester quadruple serum screen.

The aim of pregnancy screening for Down's syndrome is to offer women, during pregnancy, a screening test which can identify those women with an increased risk of having a child with Down's syndrome. The group at increased risk (above the defined cut-off level) can then be offered diagnostic tests and with high quality counselling, parents can then make informed choices. The definitive diagnostic tests offered require either amniocentesis or chorionic villus sampling. These are invasive procedures which carry a small, but significant, risk of fetal loss.

As the screening performance has significantly improved with the enhancements previously set out in CEL 31(2008) the cut-off for both first and second trimester screens can be set at 1 in 150 at term without any substantial loss in detection rate. This will assist in meeting the NSC's current standard which requires the programme to achieve a detection rate (DR) of greater than 90% of affected pregnancies with a screen positive rate (SPR) of less than 2% (of affected pregnancies) for the combined first trimester screen and a detection rate (DR) of more than 75%, for a screen positive rate (SPR) of less than 3% (of affected pregnancies) for second trimester quadruple screening. As a result, this should mean fewer diagnostic tests are therefore needed, leading to a reduction in the rate of loss of unaffected fetuses.

From a laboratory perspective, the four markers used in the second trimester screen perform better at slightly different gestations. Thus, in striking a balance between the benefits of all the markers, second trimester quadruple screening can be offered from 14 weeks + 2 days to 20 weeks + 0 days. This means that there will be a seamless transition between the combined and quadruple screen that can be offered for Down's syndrome. Any woman presenting for the first trimester screen who is subsequently found to be beyond the gestational range can immediately be offered quadruple screening as an alternative and they will not need to return for a further appointment.

It has to be acknowledged that the second trimester test recommended is not as sensitive and specific as the first trimester screen. Therefore women should be encouraged to present early and be made aware of the benefits of doing so if they wish screening.

In regard to reporting a risk for Trisomy 13 (Patau's syndrome) and Trisomy 18 (Edwards' syndrome), current evidence does not support introducing tests for these conditions in the first trimester at present as a national policy as some aspects do not meet the UK NSC criteria for screening. An expert group, with appropriate Scottish representation, will be established to review the evidence through the UK NSC review process.