

Dear Colleague

CHANGES TO THE PREGNANCY AND NEWBORN SCREENING PROGRAMMES

The annex to this letter sets out a number of changes and developments to strengthen and extend the pregnancy and newborn screening programmes. These changes take account of the updated advice from the UK NSC and the recommendations of the NHS QIS Health Technology Assessment Report 5 – *Routine Ultrasound Scanning before 24 Weeks of Pregnancy*.

In summary, these changes are:

1. The replacement of the existing Pregnancy Screening Programme offered for Down's Syndrome and other congenital anomalies
2. The introduction of haemoglobinopathy screening both during pregnancy and for newborn babies
3. The extension of the newborn bloodspot screening programme to include screening for Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD).

NHS Boards will be responsible for implementing the changes in maternity and child health services required to deliver these improvements no later than 31 March 2011. NHS Boards should also be offering as routine a fetal anomaly scan by the end of 2009. Support from the National Services Division (NSD) will be available. A breakdown of individual Board funding is at Annex A.

NHS Quality Improvement Scotland to revise and review the clinical standards for pregnancy and newborn screening.

NHS Health Scotland to develop and produce the required patient information material and health professionals training materials

NHS Education Scotland to work with NSD to coordinate and lead the education and training of health professionals involved in the pregnancy and newborn screening programmes.

CEL 31 (2008)

July 2008

Addresses

For action

Chief Executive, NHS
Boards
Directors of Public Health

For information

Royal College of general
Practitioners
Royal College of Nursing,
Scottish Division
Royal College of Midwives
General Medical Council
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Screening Enquires to:

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NHS National Services Scotland, National Services Division (NSD) to lead, co-ordinate and support NHS Boards in making the changes. Additional funding will be allocated for:

- Implementation of the development required in the pregnancy screening laboratories.
- Commissioning the Scottish newborn screening laboratory to introduce sickle cell and MCADD screening into the newborn bloodspot screening programme.

A final decision on the rationalisation of the laboratory service has yet to be made. Further advice on this issue will follow later in the year.

Health Inequalities and Joined-up Working

The commitment to tackle health inequalities is a priority. **equally well: report of the ministerial task force on health inequalities** was published in June and recommends that:

- *NHS Boards should improve the capacity of ante-natal services to reach higher risk groups and identify and manage risks during pregnancy.*

NHS Boards are expected to ensure services are redesigned to maximise the opportunity for women who are disadvantaged to have access to the best possible screening service and ante-natal care.

Finally, **Healthy Eating Active Living, an action plan to improve diet, increase physical activity and tackle obesity (2008-2011)** includes a commitment to improve the nutrition of pregnant women and women of child bearing age as well as to support an increase in the uptake of Healthy Start. Delivery will be supported by significant resource over the next three years. A more specific CEL will be issued in the next few weeks. But the changes in screening services present an opportunity to improve the co-ordination of activities aimed at pregnant women.

Yours sincerely



PAM WHITTLE

Director of Public Health and Wellbeing

CHANGES TO PREGNANCY AND NEWBORN SCREENING PROGRAMMES

Pregnancy Screening for Down's Syndrome

All pregnant women in Scotland are currently offered a serum screening programme, which includes the measurement of 2 biochemical markers in the second trimester of pregnancy. Evidence shows that a more robust risk estimation for the pregnancy can be obtained by offering a screening programme 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 (Combined Ultrasound and Biochemical Screening (CUBS)).

Down's syndrome and some other congenital malformations are associated with an increase in the size of this fluid filled space at the back of the neck. Introduction of combined first trimester screening will enable the programme to meet the NSC's current standard which requires the programme to achieve a 75% detection rate with less than 3% false positive rate. The standard for 2010 and beyond is expected to be more stringent. Establishment of first trimester screening in Scotland, however, will put in place a framework which is sufficiently flexible to incorporate any changes that may be required to implement further scientific developments and to obtain the higher standards in the future.

For those women who do not present early enough in their pregnancy to take advantage of first trimester screening, the second trimester serum screening programme that will be offered to them, will be strengthened by the addition of measurements of 2 additional biochemical markers (quadruple test) which will refine the risk assessment available to them.

All women are also to receive a second trimester fetal anomaly ultrasound scan between 18 weeks, 0 days and 20 weeks, 6 days.

NHS Boards - 2008 to 2011 allocations

A one-off revenue allocation of £4.60M will be made in 2008-09, together with £1.65M of recurrent revenue funding in 2008-09 and the subsequent 2 financial years. This will allow Boards to purchase ultrasound machines that meet the required UK NSC specification for the Nuchal Translucency scan.

Where planned expenditure is capital in nature a transfer from revenue budgets to capital budgets is possible. Notification of such a requirement should be made to Phyllis Haggarty (Phyllis.haggarty@scotland.gsi.gov.uk) or Mike Baxter (mike.baxter@scotland.gsi.gov.uk) within the Private Finance and Capital Unit no later than end of November in this financial year.

ANNEX A

NHS Board	2008/09 One-off Revenue Allocation (£)	2008/09 Recurrent Revenue Allocation (£)	2009/10 Recurrent Revenue Allocation (£)	2010/11 Recurrent Revenue Allocation (£)
Ayrshire and Arran	348,506.01	125,157.13	125,157.13	125,157.13
Borders	154,480.44	54,105.51	54,105.51	54,105.51
Dumfries and Galloway	189,120.36	66,790.55	66,790.55	66,790.55
Fife	315,024.84	112,896.42	112,896.42	112,896.42
Forth Valley	262,875.09	93,799.33	93,799.33	93,799.33
Grampian	398,357.77	143,412.71	143,412.71	143,412.71
Greater Glasgow	971,030.40	353,123.81	353,123.81	353,123.81
Highland	304,955.82	109,209.17	109,209.17	109,209.17
Lanarkshire	455,490.75	164,334.64	164,334.64	164,334.64
Lothian	556,831.83	201,445.46	201,445.46	201,445.46
Orkney	89,839.60	30,434.22	30,434.22	30,434.22
Shetland	92,475.81	31,399.59	31,399.59	31,399.59
Tayside	358,398.91	128,779.88	128,779.88	128,779.88
Western Isles	102,612.37	35,111.57	35,111.57	35,111.57
Totals	4,600,000	1,650,000	1,650,000	1,650,000

Haemoglobinopathies

The haemoglobinopathies are a large group of inherited blood disorders which affect the haemoglobin (oxygen carrying) component of blood. They fall into two main groups – the haemoglobin variants (such as sickle cell disorders) which are associated with the production of abnormal forms of haemoglobin, and the thalassaemias in which there is an abnormality in the amount of haemoglobin produced. Many haemoglobinopathies are of no clinical significance whereas others are associated with severe morbidity and mortality, most notably sickle cell disorders and beta thalassaemia major. Sickle cell disorders, caused by a variant haemoglobin, often result in severe life threatening clinical symptoms. Those with beta thalassaemia major require regular blood transfusions to maintain life.

Though no haemoglobinopathy is exclusive to any single ethnic group, the frequency of these disorders varies considerably in different ethnic groups. These disorders originated in areas of the world where malaria is, or was, endemic because their occurrence conferred a survival advantage to those living in such areas. Thus, though haemoglobinopathies may be encountered in northern Europe, they are mainly associated with populations whose ancestry originated in Africa, Asia or around the Mediterranean.

The aim of offering screening in the antenatal period is to identify couples who are at risk of having an affected child and thereby offer them information on which to base reproductive choices. It is important that screening is offered early so that the results of the screening tests and any prenatal diagnosis are available sufficiently early for couples to be able to make timely informed choices. Screening for sickle cell disorders and thalassaemia should be offered to all women as early as possible in pregnancy, and ideally by 10 weeks.

In respect to newborn babies, neonatal screening is intended to identify newborns that are affected with sickle cell disorders in order to promptly institute penicillin prophylaxis and comprehensive care which has been shown to reduce morbidity and mortality.

Haemoglobinopathy screening will be introduced by:

- Offering all pregnant women screening for thalassaemia based on a formal process of inspection of routine blood indices
- Offering women in high risk groups, or women whose partners are in high risk groups, screening for sickle cell disorders and other haemoglobin variants using a recommended Family Origin (Ancestry) Questionnaire to assess risk status.
- Adding universal screening for sickle cell disorders to the newborn bloodspot screening programme for a trial period.
- Linking the pregnancy and newborn haemoglobinopathy screening programmes in an 18 month pilot project to determine if cases would be missed if a policy of targeted screening of newborns was adopted.

Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD)

MCADD is an inherited metabolic disorder which occurs with roughly the same incidence as PKU, for which newborn babies are already offered screening. The abnormality leads to an inability to metabolise sufficient energy from fat during periods of stress such as fasting, inter current illnesses with fever or surgery. It is a recognised cause of unexpected death in infancy and of acute encephalopathy in infancy requiring intensive care, which has significant subsequent morbidity and mortality. Although rare, a significant proportion of individuals with MCADD die or have serious longer term outcomes.

In a recent large pilot study in England in which over 800,000 babies were screened, a significant reduction in mortality was shown by the implementation of relatively straight forward interventions when cases were detected. Early recognition allows the introduction of appropriate feeding regimes, which can be supplemented during periods of stress, as well as early implementation of appropriate management should the child require hospitalisation.

This screening will be introduced by the extension of the newborn bloodspot screening programme.