UK National Screening Committee (UK NSC)
Note of the meeting held on 25 April 2012
at
Royal Free Hampstead NHS Trust
344 - 354 Gray's Inn Road
London WC1X 8BP

Present
Dr Gordon Paterson (Chair)
Ms Alex Berry
Dr Sunil Bhanot
Dr Margaret Boyle
Professor Roger Brownsword
Professor Martin Buxton
Ms Majella Byrne
Dr Catherine Calderwood
Professor Alan Cameron
Dr Jennie Carpenter
Dr David Elliman
Ms Jane Fisher
Dr Rosemary Fox
Dr Nick Hicks
Mrs Madeleine Johnson
Dr Surendra Kumar
Dr Janet Little
Dr Anne Mackie
Ms Cheryl Paris
Professor Julietta Patnick
Mr Nick Waddell
Dr Jane Wilkinson

Visitors
Ms Diane Dempster
Mr Tim Elliott
Mr John Marshall
Professor Catherine Peckham
Dr Allison Streetly

Secretariat
Miss Jo Taylor
Miss Kathryn Flynn

Apologies
Sir Harry Burns
Mrs Clare Brassington
Professor Gareth Evans
Mrs Moira Morris
Dr David Walker

Welcome and Introductions

1.0 Dr Gordon Paterson had agreed to chair the meeting in Sir Harry Burns's absence. Dr Paterson welcomed all to the meeting including:-

New UK NSC Member

- Ms Alex Berry, Divisional Director for South East London, London Specialised Commissioning. Ms Berry will be giving advice to the committee on specialised commissioning in England.

New Observer

- Ms Majella Byrne, Acting Director National Cancer Screening Service, who will be replacing Dr Alan Smith as the Republic of Ireland's observer on the UK NSC.

Agenda Item Presenters

- Mr Tim Elliott, Cancer Policy Team, Department of Health presenting the bowel and breast cancer screening agenda items.
- Mr John Marshall, UK NSC Projects and Programmes Manager presenting the agenda items on asymptomatic bacteriuria screening in pregnancy with Professor Catherine Peckham and the newborn screening for Duchenne muscular dystrophy agenda item with Dr Anne Mackie.
- Professor Catherine Peckham CBE, Programme Director, NHS Infectious Diseases in Pregnancy Screening Programme presenting the agenda item on rubella susceptibility screening in pregnancy, the asymptomatic bacteriuria screening in pregnancy agenda item with Mr John Marshall and the screening for cytomegalovirus agenda item.
- Dr Allison Streetly, Programme Director, NHS Sickle Cell and Thalassaemia Screening Programme presenting the agenda item on newborn thalassaemia screening with Dr Anne Mackie.

Observers

- Ms Diane Dempster, Scottish Government.

Resignations

1.1 The Chair informed members that Dr Lesley Wilkie has retired from her post as Director of Public Health for NHS Grampian and has therefore resigned from the committee. Members asked the Chair to formally write to Dr Wilkie to thank her for her personal contribution to the work of the committee over the years. The secretariat were seeking a new member to replace Dr Wilkie.

Vacancies

1.2 Miss Josephine Taylor stated that there had been a vacancy on the committee for a consumer organisation representative for sometime and asked members to send her any nominations for the vacancy.

Action: Members to send any nominations to Miss Josephine Taylor

2.0 Minutes and Matters arising

2.1 The minutes of the last meeting were agreed, subject to amending Dr Rosemary Fox's job title to Director of Screening Services and a minor typographical error on page three.

2.2 There were eight actions points from the last meeting:-

3.5 Update on Cost-effectiveness Paper

A paper on cost-effectiveness will be brought to the UK NSC in 12-18 month's time as by then Public Health England will have been established and the National Institute for Health and Clinical Excellence (NICE) will have also reviewed their methodology on cost effectiveness, therefore this is an ongoing action point.

4.37 Fragile X Syndrome Screening in Pregnancy Policy Position Statement

Mr John Marshall has amended the title of the Fragile X syndrome screening review. A copy of the review is available at http://www.screening.nhs.uk/fragilex

4.40 Fragile X Syndrome Screening in Pregnancy Policy Position Statement

Dr Anne Mackie has written to the Clinical Genetics Society about the UK NSC's recommendation on Fragile X syndrome screening in pregnancy and to highlight the need to review advice on test cut offs and the possibility of undertaking an assessment of current cascade testing practices in the UK.

5.6 Update to the Handbook for Vascular Risk Assessment, Risk Reduction and Risk Management

Dr Mackie has amended the Vascular Handbook to reflect member's comments.

5.28 Screening for Familial Hypercholesterolaemia Policy Position Statement

Dr Mackie has written to the NHS Commissioning Board Authority about cascade screening for familial hypercholesterolaemia.

5.36 Draft policy for age of first screen for cervical abnormalities and frequency of screening for those aged 50-64 years

Members have sent comments to Dr Mackie on the draft age of first screen for cervical abnormalities and frequency of screening for those aged 50-64 years paper. A copy of the revised paper will be placed on the UK NSC website for consultation shortly.

5.40 and 5.41 Draft policy for Human Papilloma Virus testing to alter follow up regimes in cervical cancer

The evaluation of the sentinel sites is included in the meeting papers and Human Papilloma Virus testing to alter follow up regimes in cervical cancer paper is for discussion at today's meeting.

3.0 Director's Report Back

3.1 Dr Mackie gave an update as follows:-

Update on Newborn Screening for Maple Syrup Urine Disease (MSUD), Homocystinuria (pyridoxine unresponsive), Glutaric Aciduria Type I (GA1), Isovaleric Acidaemia (IVA) and Long-chain 3 - hydroxyacyl CoA dehydrogenase deficiency (LCHADD)

3.2 Dr Mackie said that the evaluation to investigate whether there is evidence to support including the above five conditions to the newborn bloodspot screening programme would begin in July 2012. The evaluation will last for one year and will be funded and run by the National Institute for Health Research and will cover the populations served by the screening laboratories in Leeds, Manchester, Sheffield, Birmingham, London (Guy's and St Thomas' and Great Ormond Street). It is estimated that 430,000 babies will be tested for the conditions. A steering group has been set up to monitor the evaluation. Dr Mackie stated that the results of the evaluation will be brought to the Autumn 2013 meeting of the UK NSC.

Preconception Testing Statement

3.3 Dr Mackie said that implementation of the NHS Sickle Cell and Thalassaemia (SC&T) Screening Programme has raised awareness of the conditions and led to many couples seeking advice and testing, for their own carrier status outside of the antenatal period and had highlighted the need for guidance on this matter. Dr Mackie stated that the NHS SC&T Screening Programme has produced guidance on this following a request from colleagues working within primary care. The guidance which is jointly agreed with the Joint Committee of Medical Genetics (JCMG) suggests that it is appropriate to offer testing to couples at higher risk or who believe they might be at higher risk, following appropriate advice and counselling. It has been shared with the Academy of Medical Royal Colleges. Dr Streetly reported that the JCMG recommended to the Royal Colleges that they contact the NHS SC&T Screening Programme if

they would like further guidance to ensure that adequate information and knowledge is available to healthcare professionals.

4.0 Adult Screening

4.1 The Chair explained that the normal format for the meeting had been changed to accommodate speakers' other commitments therefore the adult screening agenda items would be discussed first.

Update on Abdominal Aortic Aneurysm Screening

4.2 Dr Mackie said that the national implementation of the NHS Abdominal Aortic Aneurysm (AAA) Screening Programme remains on track to cover the whole of England by March 2013. Phase 3, the largest phase of national implementation, is nearing completion. Dr Mackie stated that the roll-out of the Phase 3 programmes was officially launched by the Secretary of State for Health in October 2011. The remaining 25% of the country will implement screening during Phase 4 between October 2012 and March 2013.

4.3 Dr Mackie stated that the first ever UK wide audit of elective AAA repair surgery was published on 29 February. The audit showed that mortality rates for elective AAA repairs fell by as much as two thirds in just four years between 2006 and 2010 (from 7.5 per cent to 2.4 per cent). The audit showed that all local AAA screening programmes that were screening during the reporting period (October 2008 to September 2010) met the NHS AAA Screening Programme's quality standard for a postoperative mortality rate of less than 6% for elective AAA repair surgery.

4.4 Members asked about the online patient decision aid for AAA screening that is being developed as part of the National Shared Decision Making Programme to support men and their families in making an informed decision about AAA screening. Mr Nick Waddell said that the first stage of development was complete and it is expected that it will be ready for testing this summer ahead of its launch later in the year.

4.5 Dr Jane Wilkinson said that planning for the introduction of an AAA screening programme in Wales is well underway. The full programme costs are yet to be secured therefore the implementation date cannot be confirmed.

4.6 Dr Margaret Boyle said that AAA screening will be implemented across Northern Ireland in June 2012. It will be delivered in a number of health and social care locations across Northern Ireland.

4.7 Ms Cheryl Paris said that the Scottish AAA Screening Programme will commence implementation in June 2012 with national coverage of the programme expected by October 2013.

4.8 The UK NSC noted the position in each of the four UK countries.

Cervical Screening

4.9 Dr Mackie said there were three different strands of work ongoing with cervical screening. These are:-

Draft policy for age of first screen for cervical abnormalities and frequency of screening for those aged 50-64 years

4.10 At the UK NSC meeting on 17th November 2011, Dr Mackie asked members to send her comments on the draft policy for age of first screen for cervical abnormalities and frequency of screening for those aged 50-64 years paper. Dr Mackie stated that these comments had now been included within the paper and the paper should be available on the UK NSC website for consultation within the next couple of weeks.

Action: Dr Mackie to place the draft policy for age of first screen for cervical abnormalities and frequency of screening for those aged 50-64 years on the UK NSC website for consultation

Draft policy for HPV testing to alter follow up regimes in cervical cancer

4.11 Dr Mackie said at the UK NSC meeting on 17th November 2011 members had discussed the use of Human Papilloma Virus (HPV) testing as "triage" in cervical screening and as a "test of cure" for women previously treated for cervical abnormalities.

4.12 A HPV pilot scheme, which completed in 2006, concluded that HPV testing could be beneficial for the triage of women with low grade cervical abnormalities. Based on this evidence, the Sentinel Site Implementation Project examined the practical implementation of any national roll-out of HPV testing, including using HPV testing as a "test of cure" for women previously treated for cervical abnormalities. Following a request at the UK NSC meeting in November a copy of the Sentinel Sites evaluation had been circulated to members. The UK NSC noted the evaluation and agreed that the draft policy they had discussed at November's meeting should be placed on the UK NSC website for consultation.

Action: Dr Mackie to place the draft policy for HPV testing to alter follow up regimes in cervical cancer paper on the UK NSC website for consultation

HPV as a Primary Screen for Cervical Cancer

4.13 Professor Julietta Patnick said that a feasibility study was currently looking at whether HPV testing could be used as a primary screen for cervical abnormalities. The Advisory Committee on Cervical Screening in England had set up three working groups to inform the potential study on: workforce requirements and supporting transition; protocols and algorithms for testing; and building the business case to demonstrate the economies and benefits to women of HPV Testing as Primary Screening (HPV TaPS). Professor Patnick

said that all available data suggested that HPV TaPS does indeed result in fewer cases of cervical cancer on follow-up.

4.14 Members asked about self-testing. Professor Patnick said that the STRATEGIC trial to increase cervical screening uptake at first invitation study is exploring this (STRATEGIes to increase Cervical screening uptake at first invitation). More information on the study is available at: http://www.controlled-trials.com/ISRCTN52303479

4.15 Members also asked about the cost-effectiveness of HPV TaPS. Professor Patnick said the ARTISTIC trial had looked at both clinical and cost-effectiveness but further modelling would be needed as part of the feasibility study.

4.16 The UK NSC agreed that there is enough evidence to suggest that HPV TaPS would be cost and clinically effective. It was agreed that the UK NSC should consult on a recommendation to approve HPV as a primary screen for cervical cancer and that the feasibility study should explore implementation issues including length of time before a re-screen following a HPV negative result.

Action: Dr Mackie to place the draft policy for HPV as Primary Screen for Cervical Cancer paper on the UK NSC website for consultation

Prostate Cancer Screening

4.17 Dr Mackie said that in March 2012 an update of the European Randomized Study on Screening for Prostate Cancer (ERSPC) study results were published which reports primary outcome measures for 11 years of follow-up as compared to the median 9 years follow–up in the 2009 publication from the same trial. The data was published in the New England Journal of Medicine: http://www.nejm.org/doi/full/10.1056/NEJMoa1113135

4.18 Dr Mackie asked the School of Health and Related Research (ScHARR) to examine the findings and to see how the data compared to the data included within their model produced for the UK NSC in 2009. ScHARR reported that the headline data matched their model. However, the prostate cancer survival estimated from cumulative hazard data suggested a significant improvement. As there were discrepancies in the figures Dr Mackie had contacted the authors of the study, who confirmed there was an error in the printed paper. The range on the cumulative hazard of death from prostate cancer graph (y axis) should have been from 0 to 0.014 not 0.14 as was printed.

4.19 Dr Mackie said she would ask ScHARR to recalibrate their model on prostate cancer screening using the revised figures. However, it was thought that the revised figures were unlikely to impact on the conclusions drawn from the previous UK NSC review of prostate cancer screening.

4.20 Mr Tim Elliott said that the Riskman Trial set up by the Prostate Cancer Support Federation, the University of Warwick, and the Institute of Cancer Research aims to show that screening using additional information, such as

family history, ethnicity, age, and digital rectal examination, will be a better marker at predicting prostate cancer than the Prostate Specific Antigen (PSA) test alone. This will mean that a number of men will not have to undergo an unnecessary biopsy, with all the potential anxiety, discomfort and risk of infection that holds. Mr Elliott said that the results from this trial could change the dynamics of a potential national screening programme, but there were issues around securing funding for the trial. Mr Elliott said he would keep members updated on the trial.

4.21 The UK NSC noted the updated figures.

Action: Dr Mackie to ask ScHARR to recalibrate the model on prostate cancer screening and to circulate a revised paper to members

Bowel Cancer Screening

4.22 Professor Patnick said that plans to extend the NHS Bowel Cancer Screening Programme in England to screen people up to the age of 75 using the faecal occult blood test (FOBT) were continuing. However, the age extension of the original FOBT programme is stalling in some areas due to issues around endoscopy capacity. A cross Department of Health and NHS group has been set up to address this, and NHS Improvement is due to begin working with a number of sites to help streamline their endoscopy services and make the most use of capacity.

4.23 Professor Patnick said that advice and bidding documentation on the flexible sigmoidoscopy (FS) pilot was issued to the NHS in January 2012. Strategic Health Authority (SHA) clusters were asked to nominate potential pilot sites amongst their local screening centres against a set of strict criteria. The pilots are due to be selected later in April. SHA clusters were also asked to nominate first wave of roll-out sites at the same time. The pilot sites are due to begin screening in the autumn.

4.24 Members asked whether there had been any progress in improving uptake rates. Mr Tim Elliott said it was hoped the first national NHS campaign to raise awareness of the signs and symptoms of bowel cancer would reach those people who do not usually accept the offer of screening. The Be Clear On Cancer Campaign included TV, radio, press, bus and online advertising, as well as a series of events across England. The campaign would be evaluated and the findings would be shared with members later in the year.

Action: Mr Tim Elliott to bring the evaluation to a future meeting

4.25 Dr Jane Wilkinson said Bowel Screening Wales invites both men and women aged 60-74 for bowel screening every two years using FOBT. Uptake rates are approximately 54-55%.

4.26 Ms Cheryl Paris said the Scottish Bowel Screening Programme invites men and women in Scotland between the ages of 50 to 74 for screening every two years. Ms Paris said Scotland are looking at running a study for younger men

and women using FS or the faecal immunochemical test (FIT). The findings of which will be shared with the UK NSC.

4.27 Dr Margaret Boyle said the Northern Ireland Bowel Cancer Screening Programme is offered to both men and women aged 60-71 every 2 years using FOBT. Uptake rates are approximately 50%.

4.28 All three countries said there were issues around endoscopy capacity.

4.29 Ms Majella Byrne said the Republic of Ireland is currently preparing for the introduction of a national bowel cancer screening programme, for men and women aged 55 to 74 using FIT. The screening programme will be implemented on a phased basis starting with men and women aged 60-69. Over time the programme will be extended on a phased basis until the full 55-74 age group is reached.

4.30 Dr Mackie said that an evaluation of FIT against the UK NSC criteria should be brought to a future UK NSC meeting.

Action: Dr Mackie to bring an evaluation of FIT against the UK NSC criteria to a future meeting

Update on Breast Screening Review in England

4.31 Mr Elliott said the independent review was ongoing and it is envisaged it will report by the summer. A copy of the review will be brought to a future UK NSC meeting.

Action: Mr Tim Elliott to bring a copy of the Breast Screening Review in England to a future meeting

5.0 Fetal Maternal and Child Health Screening

Report from Fetal, Maternal and Child Health Co-ordinating Group

5.1 Mrs Madeleine Johnson, Chair of the Fetal, Maternal and Child Health Co-ordinating Group (FMCH) said that there had been two meetings of the FMCH since the UK NSC had last met. These took place in November and March. Mrs Johnson reported that:-

Policy Reviews

5.2 The FMCH had discussed screening reviews for Duchenne muscular dystrophy, rubella susceptibility, cytomegalovirus, asymptomatic bacteriuria and newborn thalassaemia screening. These policy reviews are main agenda items for the UK NSC meeting.

5.3 Mrs Johnson said that consultations for the following conditions are currently open or due to open imminently HTLV, adolescent idiopathic scoliosis,

kernicterus, Tay Sachs disease, Canavan disease, familial dysautonomia and antenatal screening for feto-maternal alloimmune thrombocytopenia.

5.4 Reviews due for consideration at the next FMCH meeting are severe combined immunodeficiency, iron deficiency anaemia in childhood, biliary atresia, biotidinase deficiency and group B streptococcal carriage in pregnancy.

First trimester screening for Trisomy13 (T13) / Trisomy 18 (T18)

5.5 Mrs Johnson stated that screening currently takes place during the 18 – 20 week scan, however, about a quarter of laboratories were reporting results for these conditions in the first trimester as an incidental finding from Down's syndrome screening. Establishing an algorithm combining measurement of nuchal translucency and biochemical markers could increase first trimester detection. The FMCH had agreed that this should be evaluated against the UK NSC criteria.

Repeat screening for congenital hypothyroidism (CHT) in pre-term infants

5.6 Mrs Johnson said that the current newborn screening policy for CHT recommended that all babies born at less than 36 completed weeks should have a repeat test at the equivalent of 36 weeks gestation. However, this policy proved complex and difficult to implement. Following debate within the Newborn Bloodspot Screening Programme and consultation with stakeholders it was proposed that:

'All babies whose gestational age is less than 32 completed weeks (31+6 days) would have repeat testing at 28 days postnatal age (counting the date of birth as day 0), or at the time of being discharged home, whichever is the sooner.'

5.7 The FMCH endorsed the revised protocol and it was agreed to report it to the UK NSC.

5.8 The UK NSC noted the FMCH update.

Rubella Susceptibility Screening in Pregnancy Policy Position Statement

5.9 Dr Surendra Kumar asked if it could be recorded that he had chaired the General Medical Council Panel which investigated research into links between autism and the MMR vaccine.

5.10 Professor Catherine Peckham said that rubella screening was offered before the creation of the UK NSC. When the policy for screening for rubella susceptibility was last reviewed by the UK NSC in 2003 it did not meet its criteria. However, a pragmatic decision was made that screening should continue because of the declining MMR rates.

5.11 The current review again recommends that screening for rubella susceptibility does not meet the UK NSC criteria. This is because of:

- the low incidence of congenital rubella syndrome
- screening in pregnancy does not contribute to the reduction of risk in the current pregnancy
- the test may falsely reassure some women that they are not susceptible to rubella infection
- screening and immunisation of postnatal women may not be the optimum intervention to address rubella susceptibility as a public health issue in the whole adult population
- the opportunity cost of improving the programme may outweigh the benefits to be derived from doing so

5.12 Professor Peckham said that the Joint Committee on Vaccination and Immunisation (JCVI) supported this policy. Since the consultation closed a meeting has taken place with the Department of Health in England's Immunisation Branch and the Health Protection Agency (HPA). It was agreed that work should be undertaken to establish an overall direction of travel towards:

- selection of postnatal vaccination population through vaccination history rather than antenatal screening
- inclusion of MMR in ongoing JCVI work to strengthen the current immunisation catch up policy in adolescence

5.13 The UK NSC agreed the policy position on rubella susceptibility screening in pregnancy as screening for rubella susceptibility does not meet the UK NSC criteria for a screening programme. The UK has a very low incidence of congenital rubella syndrome but increasing reports of susceptibility in pregnant women. In this context there is concern that the current approach based on screening all pregnant women and vaccinating those found to be susceptible is not the optimum approach to preventing outbreaks of rubella infection.

5.14 The UK NSC agreed that the present arrangements for antenatal screening and post partum immunisation should continue until other arrangements are in place and that the policy should be reviewed in three years' time unless there is significant new peer reviewed evidence in the meantime.

Asymptomatic Bacteriuria Screening in Pregnancy Policy Position Statement

5.15 Mr John Marshall said that screening for asymptomatic bacteriuria in early pregnancy is an established part of antenatal care packages and was introduced before the UK NSC was created. The current policy is that screening for this condition should be offered. However, a systematic population screening programme is not recommended as clinical practice guidelines are covered by NICE. This policy was adopted following discussion within the UK NSC's antenatal subgroup in 2004.

5.16 The recommendation in 2004 was based on clinical guidelines published in 2003 where the purpose of screening and treating women with positive results

was to prevent preterm labour. More recent guidelines published by NICE in 2008 state that the purpose of screening and treating women with positive results is to prevent pyelonephritis in pregnancy.

5.17 This change followed two major publications:

- the HTA study, 'Screening to prevent spontaneous preterm birth: systematic reviews of accuracy and effectiveness literature with economic modelling' (2009). This study estimated that 30% of women with asymptomatic bacteriuria will develop pyelonephritis if left untreated. The study also found that a positive test had a poor predictive value for preterm labour but that antibiotic treatment was effective in preventing preterm labour.

- the updated Cochrane review, 'Antibiotics for asymptomatic bacteriuria in pregnancy' (2007). This review found no clear evidence for an association between the condition, asymptomatic bacteriuria, and the outcome, preterm labour. Neither did it find evidence that antibiotic treatment reduced preterm labour. The review did find that antibiotic treatment was effective in clearing asymptomatic bacteriuria and in reducing pyelonephritis and low birth weight.

5.18 The screening review suggested that, while there is value in continuing to recommend screening, there is insufficient information to recommend a population screening programme. The key knowledge gaps relate to the current prevalence of asymptomatic bacteriuria, the impact of screening on pyelonephritis as a whole, the optimum test, its timing and frequency during the pregnancy and the optimum treatment strategy.

5.19 The UK NSC agreed its policy position as testing for asymptomatic bacteriuria should be offered as part of routine clinical management and agreed that the policy should be reviewed in three years' time unless there is significant new peer reviewed evidence in the meantime.

5.20 The committee also agreed to the recommendation that guideline development in this area should consider the issues identified by the review. It was agreed that the HPA should be contacted about the current prevalence rates of asymptomatic bacteriuria.

Action: Mr John Marshall to write to NICE and the HPA about the issues identified in the asymptomatic bacteriuria screening in pregnancy review

Newborn Screening for Thalassaemia Policy Position Statement

5.21 Dr Allison Streetly said that the policy position since the introduction of newborn screening for sickle cell disease in 2001 has been that β Thalassaemia Major does not meet the criteria for the introduction of a screening programme. However, there have been some important developments since the last review. Concerns have been expressed about waiting for babies to present with thalassaemia when they become unwell

rather that identifying them through screening. Some thalassaemias are picked up routinely as incidental or by-product finding of the newborn sickle cell screening programme and some babies are picked up through antenatal testing. There have also been developments in treatment since the screening programme was started in 2001 (oral and cardio-protective iron chelators) which have improved outcomes and long term survival of adults. More recently evidence on the continuing improved treatment and survival in Europe compared to the USA (which did not licence some drugs until recently) has been reported.

5.22 Dr Streetly said there has been on-going "noise" in the system about the approach to newborn screening for thalassaemia and a perceived lack of clarity about these cases. As a result of these issues a report reviewing the case for newborn screening was commissioned from Solutions for Public Health. The published evidence relating to four thalassaemias was considered, the most significant being thalassaemia major. The report recommends that a newborn screening programme should not be introduced for β-thalassaemia intermedia and Hb E/ β-thalassaemia or for Hb H disease. In the context of limited published evidence, the report identified a need for a randomised controlled trial in the case of β-thalassaemia intermedia and E/ β-thalassaemia to allow more information to be gathered but recognised that a trial of early versus late treatment of β-thalassaemia major would be unethical. The report did not reach a conclusion on the evidence of screening for β thalassaemia major.

5.23 The report also considered it was necessary to establish ways of managing the incidental findings of thalassaemias generated by the newborn programme It was suggested that the NHS SC&T Screening Programme should develop patient and professional information relating to reporting and managing clinically significant thalassaemias including ensuring information is available for parents which informs them that these conditions may be found on the newborn bloodspot test.

5.24 It was also suggested that the screening programme should contribute to evaluation and research initiatives to develop the knowledge base in key areas, for example regarding the working of the relevant cut-offs.

5.25 The replies to the consultation were discussed by members. Dr Streetly said the FMCH had raised no objections to the review recommendations.

5.26 The UK NSC agreed a policy position on newborn thalassaemia screening which states that screening for thalassaemia is not recommended. The published evidence relating to four thalassaemias (ß thalassaemia major, â-thalassaemia intermedia, Hb E/ β-thalassaemia and Hb H disease) was considered, the most significant being thalassaemia major. In relation to this condition there is uncertainty on whether the current test cut off value is appropriate for the population as a whole and there is a lack of published evidence to demonstrate the additional benefit of early treatment in screen detected populations versus delayed initiation of treatment following symptomatic presentation.

5.27 The UK NSC agreed that the policy of reporting clinically significant thalassaemias, found incidentally in the course of newborn screening for sickle cell disease, should continue until more evidence is available.

5.28 The UK NSC asked the NHS SC&T Screening Programme to:

- develop good practice guidance with relevant professional groups including clinicians who do find one of these conditions, further clarify laboratory guidance on reporting incidental findings and ensure that guidance to parents on newborn screening indicates that these conditions are an incidental finding of newborn screening even if not recommended for newborn screening.
- work with clinicians and laboratories to facilitate rigorous evaluation (including RCTs where appropriate) to attempt to use the opportunity of the screening programme to obtain new knowledge regarding the working of the relevant cut-offs and to facilitate answering uncertainties regarding treatment.

5.29 The UK NSC agreed that the policy should be reviewed in three years' time unless there is significant new peer reviewed evidence in the meantime.

Screening for Duchenne Muscular Dystrophy Policy Position Statement

5.30 Mr John Marshall said that the current UK NSC policy is that newborn screening for Duchenne Muscular Dystrophy (DMD) is not recommended. The last review of newborn screening for DMD took place in 2004. This was undertaken by the former Child Health Subgroup of the UK NSC.

5.31 Screening, was until recently, undertaken in Wales. The service evolved from a pilot study which began in the 1990's and the service remained active with NHS funding following the pilot. The Welsh programme has now been withdrawn as the sustainability of DMD screening was put at risk due to problems with the supply of commercially available reagents needed to undertake the test and the withdrawal of the blood spot external quality assurance scheme and quality control material by the Center for Disease Control (CDC) in the USA. The CDC quality programme was discontinued as there were not enough participants to support a viable scheme (only four laboratories world-wide participated). Wales had been the only country in the UK to provide this service. The withdrawal of the external quality assurance scheme has meant that the newborn bloodspot screening laboratory cannot be accredited by the Clinical Pathology Accreditation (CPA) for the DMD screening test. Therefore a decision was made jointly by the Welsh Government and Cardiff & Vale University Health Board to cease offering the service.

5.32 Mr Marshall said the withdrawal of the Welsh programme raised anxiety amongst Duchenne Muscular Dystrophy charities and a meeting of the All Party Parliamentary Group on Muscular Dystrophy was held to discuss this and the UK NSC review in January 2012. An action point arising from the

meeting was that member organisations should submit comments on the review. The UK NSC discussed all the replies to the consultation.

5.33 The UK NSC agreed the policy position on newborn screening for DMD as newborn screening for DMD is not recommended because of concerns about:

- the reliability of the current test
- while there is evidence of benefit from long term steroid treatment, the optimum age at which it should be initiated remains the subject of uncertainty
- there is insufficient evidence that identifying DMD in the newborn through screening improves long term outcomes in comparison to current practice
- the evidence regarding the impact of early diagnosis on parents' subsequent reproductive decision making is conflicting.'

5.34 The UK NSC agreed that the policy should be reviewed in three years' time unless there is significant new peer reviewed evidence in the meantime.

5.35 The UK NSC agreed they should write to the All Party Parliamentary Group on Muscular Dystrophy regarding a misunderstanding about the criteria for screening programmes.

Action: Mr John Marshall to write to the All Party Parliamentary Group on Muscular Dystrophy

Screening for Cytomegalovirus Policy Position Statement

5.36 Professor Peckham said the last time the UK NSC formally considered the evidence for screening for cytomegalovirus (CMV) was in 2000. The review addressed both antenatal and newborn screening for CMV. Antenatal screening to detect women acquiring CMV for the first time in pregnancy, and newborn screening to identify infants with congenital infection. The review recommended that CMV screening should not be offered in pregnancy or neonatally.

5.37 Professor Peckham stated that when infection is acquired for the first time in pregnancy, transmission to the fetus occurs in about 30-40% of cases. Congenital CMV can also be acquired from women with previous immunity to CMV infection (recurrent infection). The majority of infants with congenital CMV infection are asymptomatic, but around 10-15% present with CMV manifestations at birth, 40-58% of whom develop adverse outcomes, including cerebral palsy, sensorineural hearing loss (SNHL) and other neurological problems. Most asymptomatic children remain unaffected, but a small proportion develop complications, most commonly hearing impairment.

5.38 Professor Alan Cameron said that testing for CMV is available abroad and he was aware of pregnant women knowing they had the infection. The UK NSC agreed that clinical management guidelines should be followed when treating those people presenting to healthcare professionals with CMV infection.

5.39 The UK NSC agreed the policy position on screening for CMV in the antenatal and neonatal period as screening for CMV in the antenatal and neonatal periods should not be offered. In pregnancy, there is uncertainty regarding natural history of primary and recurrent CMV as it relates to the risk to the fetus. The screening test for susceptibility lacks sufficient sensitivity and there is uncertainty regarding the further investigations needed to refine the risk to the fetus in women with primary infection. No interventions have been shown to be effective in preventing maternal infection or reducing the risk of transmission to the fetus. In the neonatal period, the available tests have not been shown to be sufficiently reliable for screening and there is no clear evidence of benefit from the available intravenous or oral antiviral therapies.'

5.40 The UK NSC agreed that the policy for screening for CMV in the antenatal and neonatal periods should be reviewed in three years' time unless there is significant new peer reviewed evidence in the meantime.

6.0 Updates (for information)

These are for information only.

6.1 MRC trials administered by Efficacy and Mechanism Evaluation (EME) Programme

6.2 HTA Update

6.3 SIGN Update

6.4 Healthcare Improvement Scotland Update

6.5 Journal of Medical Screening Article: Newborn screening for medium chain acyl-CoA dehydrogenase deficiency in England: prevalence, predictive value and test validity based on 1.5 million screened babies

7.0 Any Other Business

There was none.

8.0 Next Meeting

Tuesday 13th November 2012
11:30am – 3pm
Scottish Government
Edinburgh

Action Points

1. Members to send any nominations for the consumer organisation representative vacancy on the UK NSC to Miss Josephine Taylor

2. Dr Mackie to place the draft policy for age of first screen for cervical abnormalities and frequency of screening for those aged 50-64 years on the UK NSC website for consultation

3. Dr Mackie to place the draft policy paper for HPV testing to alter follow up regimes in cervical screening on the UK NSC website for consultation

4. Dr Mackie to place the draft policy paper on HPV Testing as Primary Screening for cervical abnormalities on the UK NSC website for consultation

5. Dr Mackie to ask ScHARR to recalibrate the model on prostate cancer screening and to circulate a revised paper to members

6. Mr Elliott to bring the evaluation of the Be Clear On Cancer Campaign to a future meeting

7. Dr Mackie to bring an evaluation of FIT against the UK NSC criteria to a future meeting

8. Mr Tim Elliott to bring a copy of the Breast Screening Review in England to a future meeting

9. Mr Marshall to write to NICE and the HPA about the issues identified in the asymptomatic bacteriuria screening in pregnancy review

10. Mr John Marshall to write to the All Party Parliamentary Group on Muscular Dystrophy