UK National Screening Committee (UK NSC)

Note of the meeting held on the 23 June 2017

at

The Hilton, Belfast Northern Ireland

This meeting provided recommendation on the following conditions;

- Subaneurysmal aortas in Abdominal Aortic Aneurysm (AAA)
- Thrombophilia in Pregnancy, Neonatal and Adult populations
- Tyrosinaemia Type 1
- Vasa Praevia
- Severe Combined Immunodeficiency (SCID)

Members

Professor Bob Steele	Chair
Ms Eleanor Cozens	Patient and Public Voice (PPV)
Professor Stephen Duffy	Director of the Policy Research Unit in Cancer Awareness, Screening and Early diagnosis and Professor of Cancer Screening, Centre for Cancer Prevention, Wolfson Institute of Preventive Medicine
Ms Jane Fisher	Patient and Public Voice (PPV)
Professor Chris Hyde	Public Health Specialist, University of Exeter
Dr Greg Irving	GP
Mrs Margaret Ann Powell	Patient and Public Voice
Dr Graham Shortland	Consultant Paediatrician, Cardiff and Vale University Health Board, Noah's Ark Children's Hospital for Wales and Executive Medical Director, Cardiff and Vale University Health Board, University Hospital for Wales

Observers;

Dr Hilary Angwin	Screening & Immunisation Lead, NHS England/PHE Chair of FMCH
Dr Kathryn Callaghan	Department of Health Screening Team, Emergency Preparedness and Health Protection Policy Global and Public Health Group
Dr David Elliman	Clinical lead for Newborn Infant Physical Examination and Newborn Blood Spot, PHE
Dr Ros Given – Wilson	Chair of the Adult Reference Group (ARG)
Mrs Jo Harcombe	National Lead for Stakeholder Information and Profession Education and Training
Dr Adrian Mairs	Assistant Director of Screening and Professional Issues Health and Social Care services Northern Ireland (HSCNI)
Dr Tracy Owen	Health and Social Care services Northern Ireland (HSCNI)
Dr Sue Payne	Scottish Government
Dr Heather Payne	Senior Medical Officer for Maternal and Child Health, Welsh Government
Ms Josephine Ruwende	NHS England
Karen Simpson	Department of Health, Northern Ireland

Secretariat

Dr Anne Mackie	Director of Programmes - UK National Screening Committee
Ms Zeenat Mauthoor	Secretariat
Mr John Marshall	UK NSC Evidence Lead
Dr Cristina Visintin	UK NSC Senior Evidence Review Manager

Presenters

Professor David Archard	Chair, Nuffield Council on Bioethics
Mr Jonothan Earnshaw	Vascular Surgeon and Clinical lead of the NHS Abdominal Aortic Aneurysm (AAA)
Catherine Joynson	Assistant Director- Council Projects and Inquiries, Nuffield Council of Bioethics

Apologies

Professor Roger Brownsword	School of Law, Kings College London
Professor Alan Cameron	Consultant Obstetrician at Southern General Hospital, Glasgow
Dr Paul Cross	Consultant Cellular Pathologist, Queen Elizabeth Hospital Gateshead Health NHS Foundation Trust
Dr Hilary Dobson	Consultant Radiologist and Clinical Director of the West of Scotland Breast Screening Service and Honorary Senior lecturer, University of Glasgow
Professor Gareth Evans	Consultant in Genetics Medicine, St Mary's Hospital, Manchester
Professor Alastair Gray	Director at the Health Economics Research Centre, Nuffield Department of Population Health and Professor of Health Economics at the University of Oxford
Ms Hilary Goodman	Operational Manager of Antenatal Services/Screening at Hampshire Hospitals Foundation Trust
Dr Nick Hicks	National Co-ordinating Centre for HTA
Dr Sharon Hillier	Director of Screening Division, Public Health Wales
Dr John Holden	Joint Head of Medical Division, Medical and Dental Defence Union of Scotland
Dr Anne Kilgallen	Deputy Chief Medical Officer, Northern Ireland
Ms Sarah Manson	Scottish Government
Dr Ailsa Wight	Deputy Director Emergency Preparedness and Health Protection, Department of Health

Welcome and Introductions

1. Professor Steele welcomed all to the second meeting of the year. This time hosted in Belfast. A round of introductions was initiated whilst giving members an opportunity to update the Committee on any conflicts of interest which may be relevant to this meeting. No conflicts were raised.

   Apologies were noted.

   Professor Steele congratulated Dr Graham Shortland on his appointment as vice chair of the UK NSC following an expressions of interest exercise. Dr Graham Shortland will serve as vice chair of the UK NSC for a term of three years.

   The Secretariat proposed a change to the meeting times, which allows for an earlier and slightly later finish. The Committee supported the proposal to come into effect from the October 2017 meeting.

Minutes and Matters arising

2. One correction was requested to be made to the February minutes;

   - To note Dr Anne Kilgallen's apologies at the February meeting

   Following the aforementioned correction the remainder of the minutes were confirmed as a true and accurate record. The February minutes are to be updated and uploaded as final on the UK NSC webpage.

   Ten action points were identified from the February meeting;

   (action 1) Vice Chair

   Members wishing to be considered for the role of vice chair to submit a bio to Zeenat – Completed Dr Graham Shortland has been appointed

   (action 3) Directors Update- Update of combined testing in the Fetal Anomaly Screening Programme

   Authors of Nuffield Bioethics report to be invited to the June UK NSC meeting- On the agenda

   (action 3b) Directors Update- Reference Group
   Dr Sue Payne to forward the Scottish Medical Consortium model to Zeenat- In hand

   (action 4) Annual Call for Topics

John Marshall to send out outcome letters to submitters from the Annual call for topics- Completed

(action 4b) John Marshall to confirm the 2017 date for the annual call for topics- To be announced shortly though the timeframe will be September to December for proposals to be submitted

(action 5a) Informed Choice- Task and Finish Update
Task and Finish group to update on progress at the June meeting- On the agenda

(action 5b) Progression of the group to be blogged- Completed

(action 6) Optimising Bowel cancer screening
Committee asked to review the document and forward comments to Dr Anne Mackie and Dr Sophie Whyte by 28 February for further consideration- Completed

(action 6b)Dr Anne Mackie to consider how to gain more stakeholder views on the document when the UK NSC comments had been received and considered by the ScHARR team- In hand

(action 7) FMCH

FMCH December report to be circulated to the Committee- Completed

***Director's* Update**

Dr Mackie gave an update on the following

Update on Phase 1 modelling to optimise bowel screening

3. In November 2015 the UK NSC recommended the move to using Faecal Immunochemical Testing (FIT) as the primary screen test within the NHS Bowel Cancer Screening Programme. The move has been welcomed and supported as a positive step in tackling bowel cancer as the test; only requires one sample instead of three and can detect much smaller amounts of hidden blood in the stool. It has been shown in research and pilots to increase uptake, estimated at about 10%.

3.1 Following the recommendation and announcement by the Minister, the UK NSC commissioned the Sheffield School of Health and Applied Research (ScHARR) to examine how bowel cancer screening could be optimised to use the most cost effective combination of FIT and bowel scope. The preliminary report was presented at the UK NSC's February meeting. A small workshop then followed, which brought together key stakeholders and experts to look at the model in detail. The group examined each element of the model and agreed that further work was needed.

3.2 Dr Mackie said that the comments had been shared with ScHARR who were in the process of updating the report. Further comments from the UK NSC's Adult Reference Group had also been shared along with the request for a plain English summary to be provided. Once the report had been updated Dr Mackie confirmed that this would be brought back to the UK NSC to consider further.

Action 3a: ScHARR to be invited to present the second version of the Bowel Optimisation report to the UK NSC when ready

Pulse Oximetry Report

3.3 Dr Mackie presented the UK NSC with the official summary report from the Newborn Pulse Oximetry (PO) Screening pilot. The aim of the pilot was twofold to;

- assess the impact of implementing PO in NHS Clinical services
- evaluate the feasibility of rolling this out as an additional test in the existing Newborn and Infant Physical Examination (NIPE) screening programme

3.4 The UK NSC last discussed PO at its June 2016 meeting where it agreed that a modelling exercise was required to better understand what the benefits and or or harms would be to those babies that were detected as PO screen positive but did not have critical congenital heart disease (CCHD). Dr Graham Shortland suggested that pathways and location variance be considered as part of the modelling work.

Action 3b: Dr Graham Shortland to review and input on the modelling work for PO

UK NSC Task and Finish Group to look at defining 'informed choice' and developing best practice guidance

3.5 As requested at the February meeting, the UK NSC's Task and Finish group was asked to provide an update on the group's activities.

3.6 Ms Jane Fisher reminded the Committee that the group had been brought together to address several of the Science and Technology Committee's recommendations which looked to provide an agreed definition of informed choice across the four countries and to develop a process where guidance can be developed looking at best practice and shared. Ms Fisher reported that the group had been suitably represented with positive input from the four countries, that had led to the development of the cross cutting documents presented to the Committee today.

3.7 The Committee discussed the circulated draft documents. Several points were raised on the document; the wider impact screening has not only on the individual but the impact on families, the use of the word 'personalised' in explaining informed choice and that

there is also an average "counting" age which should be factored in alongside the average reading age. Ms Fisher responded to state that in relation to the wider impact screening has this is covered by UK NSC criteria point nine under intervention. The use of the word 'personalised', was discussed at length by the group, acknowledging that some may interpret this to mean being personal to them, taking into account their individual needs and health requirements, especially in adult screening. On the other hand, some felt that the word simply reflected that an individual was making a choice that was personal to them to participate in screening. Ms Fisher said the proposed public consultation on the documents would be useful to the group.

3.8 The Committee also suggested that the explanation of 'risk' needed to be clear and be illustrated in various formats. Mrs Jo Harcombe informed the group that, PHE Screening were looking into this and would be sharing the findings with the task and finish group. From a GP perspective, Dr Irving said that GPs need more information to help in understanding risk and discussing issues with their patients. Dr Irving asked for information on why screening is not recommended to be provided about private screening. Many patients concerned about certain conditions ask why screening is not offered and note that private screening offers some where the NHS does not. Dr Mackie stated that the UK NSC has ongoing concerns with the offer of private screening from how this is advertised, offered, delivered and monitored. Dr Mackie said it was important for the UK NSC to continue to demonstrate that population screening recommended by the Committee is based on robust peer-reviewed evidence where the benefit of screening outweighs the harm. A recommended screening programme has a clear pathway and is not just a test. It supports people throughout the process from invitation to referral for treatment and advice.

3.9 The Committee agreed that the documents be put out for public consultation and be brought back to the UK NSC with the outcome. The Committee also confirmed that four country reps would confirm who would lead on supporting the guidance table once the task and finish group had ended.

Action 3c: Task and Finish group to publicly consult on guidance development documents and feedback outcome at a future UK NSC meeting

Nuffield Council on Bioethics presentation on non-invasive prenatal testing (NIPT)

4. An action following from the February UK NSC meeting was for the authors of the Nuffield report to formally present their work to the UK NSC. Catherine Joynson and Professor David Archard joined the UK NSC and to discuss their findings on the ethical, legal and regulatory implications of recent and potential future developments in non-invasive prenatal testing.

Nuffield presentation to UKNSC 23.6.17.pd

4.1 Key themes of discussion raised by the Committee consisted of: overlap of acceptability and ethics when women want screening, consideration of any legal implications, concerns around diagnostic odyssey, unknown comparative outcomes and importance of robust evidence on which screening is based.

4.2 The UK NSC thanked Ms Joynson and Professor Archard.

Adult Screening

Adult Reference Group

4.3 Dr Ros Given-Wilson, Chair of the ARG, summarised to the Committee how this group would act in a similar capacity to the Fetal Maternal and Child Health group but looking specifically at adult issues. The group would receive work from various avenues including; the regular three year review, programme modification requests and submissions via the annual call for topics.

4.4 Dr Given-Wilson confirmed that the first meeting was very productive having firstly discussed the bowel optimisation report, followed by discussion on breast screening and finally supporting the proposal for anal cancer to be deactivated from the UK NSC condition list. An evidence map developed to gauge the volume of evidence available since the last review, for anal cancer, found that there was little evidence to suggest a change in the current recommendation not to screen. Furthermore, the care of people with or at high risk of anal cancer was outside the UK NSC's remit of whole population screening and was seen more as surveillance in high risk people. The UK NSC agreed with ARG and the proposal for this to be removed from the UK NSC's active condition list. Dr Given-Wilson confirmed that stakeholders and guideline making bodies would be informed of this decision.

Screening for Subaneurysmal aortas in Abdominal Aortic Aneurysms (AAA)

5. The Chair welcomed Mr Jonothan Earnshaw to the Committee to present the proposal for screening for subaneurysmal aortas within the NHS AAA screening programme. Before commencing, the Chair highlighted to the Committee that public consultation was underway and was due to end on the 24th June.

5.1 The NHS AAA screening programme currently offers screening to 65 year old men. If their aortas measure between 3.0cms and 5.4cms they are offered surveillance as a means of monitoring the aorta's growth and managing risk. Men whose aortas measure less than 3cms are released from the screening programme with no further follow up. The proposal the UK NSC has been asked to consider is whether men who screen with an aorta measuring 2.5 to 2.9cms should enter into a lifelong surveillance programme rather than be released. Mr Earnshaw outlined that support for this proposal was to offer surveillance to more men who were borderline and may be at risk of developing an aneurysm later on in life.

5.2 A rapid review was undertaken to look at three key areas;

i. the epidemiology and natural history of subaneurysmal aortas
ii. the psychological harm associated with screening positive for AAA and/ or entering in a lifelong surveillance programme
iii. the outcomes of surgical intervention from having a subaneurysmal to an aneurysmal

5.3 Mr Earnshaw informed the Committee that whilst he supported such a proposal he agreed that there was very limited evidence to support its introduction.

5.4 Mr Earnshaw then presented to Committee a summary of the achievements the AAA screening programme had reached as the programme enters its tenth year. Future work in the programme helps to address inequalities as well as address the concern highlighted from the review on psychological harm of surveillance by developing a toolkit and developing the bespoke IT system to collect quality of life measurements.

5.5 The Committee thanked Mr Earnshaw for the presentation. They noted the ongoing public consultation and agreed to return to the issue once that had been completed.

Action 5a: UK NSC to talk to NICE about developing guidance on subaneurysmal aortas and for data to be reviewed again

NAAAASP update for NSC 2017.pptx

Fetal, Maternal and Child Health

6. Dr Hilary Angwin, Chair of the FMCH, provided the UK NSC with a verbal update following the FMCH meeting in May 2017.

6.1 The group talked though several evidence reviews in development; Dr Angwin informed the Committee that FMCH will also be considering which conditions could be removed from the main regular review cycle due to the condition no longer meeting the first criterion of being an important health problem in the UK or where the condition affects a specific sub group.

6.2 Dr Angwin confirmed that the review documents submitted to the May FMCH meeting had been approved and sent out for consultation. However the obesity review in 5 and 11 year olds required further work. Assistance from the four country reps was requested.

Action 6a: Four countries to provide Dr Elliman with information on how/if systematic measurement of child weight and height is offered in each country

UK National Screening Committee

Screening for Thrombophilia in pregnancy

7. Mr John Marshall presented the documents relating to screening for thrombophilia in pregnancy and in neonatal and adult populations to the Committee.

7.1 Thrombophilia is the term given to cover a variety of conditions where the blood clots easily. During pregnancy, women with thrombophilia are at an increased risk of developing blood clots which can cause deep vein thrombosis as well as other pregnancy complications such high blood pressure or preterm birth.

7.2 Mr Marshall informed the Committee that the evidence to screen for thrombophilia in pregnancy was last reviewed in 2010. The outcome of the 2010 review found that there was not enough evidence to support that all types of thrombophilia leads to adverse outcomes. Meaning that some women, identified as having a form of thrombophilia, would be worried unnecessarily about an outcome that may never develop. This recommendation was echoed in the findings from the TREATS health technology appraisal, which was one of the largest studies undertaken in this field.

7.3 The current review looked to see whether over the last six year any new evidence had been published to suggest that screening should be reconsidered, as well as looking at the difference between hereditary and acquired thrombophilia. The review found that there were no new studies which looked at screening in pregnancy or that which compared the outcomes of screening to no screening.

7.4 The Committee noted that only one consultation comment was received and that it was supportive of the UK NSC's recommendation not to screen for Thrombophilia.

7.5 The UK NSC discussed the review and agreed that the current recommendation not to screen for thrombophilia in pregnancy should be retained.

The Condition		
5	There should be a simple, safe, precise and valid screening test.	No studies assessing strategies for screening women was found
The intervention		

10	There should be an effective treatment or intervention for patients identified through early detection, with evidence of early treatment leading to better outcomes than late treatment.	NICE guidance available for women with risk factors
The previous 22 UK NSC criteria set were used to review the evidence. Criterion 5 refers to criterion 4 on the new 20 points UK NSC criteria Criterion 10 refers to criterion 9 on the new 20 points UK NSC criteria https://www.gov.uk/government/publications/evidence-review-criteria-national-screening-programmes/criteria-for-appraising-the-viability-effectiveness-and-appropriateness-of-a-screening-programme		

Screening for Thrombophilia in neonatal and adult populations

8. Mr John Marshall presented this item to the Committee following discussion on antenatal screening for thrombophilia. It was suggested that based on the limited evidence identified from the 2010 recommendation to not screen for thrombophilia in pregnancy, the same recommendation not to screen could be applied to neonatal and adult populations too, even though the review did not look at the evidence in these two populations.

8.1 The aim of this review was to see whether there was a sufficient volume of evidence to justify a more in-depth review to screen in the two groups and whether the imposed recommendation could be retained. As this was the first time the UK NSC was looking at screening for thrombophilia in these two groups, published evidence from 1946 to 2016 was reviewed. The review found that there was insufficient evidence relating to the two groups to answer the address key questions relating to screening strategies and effectiveness of screening.

8.2 The UK NSC recommended that based on the lack of published evidence screening for thrombophilia in neonates and adult populations should not be recommended.

The Condition		
5	There should be a simple, safe, precise and valid screening test.	No studies assessing strategies for screening in neonates and adult populations
The intervention		

10	There should be an effective treatment or intervention for patients identified through early detection, with evidence of early treatment leading to better outcomes than late treatment.	NICE guidance available for women with risk factors

The previous 22 UK NSC criteria set were used to review the evidence.

Criterion 5 refers to criterion 4 on the new 20 points UK NSC criteria
Criterion 10 refers to criterion 9 on the new 20 points UK NSC criteria
https://www.gov.uk/government/publications/evidence-review-criteria-national-screening-programmes/criteria-for-appraising-the-viability-effectiveness-and-appropriateness-of-a-screening-programme

Screening for Tyrosinaemia Type 1

9. Individuals born with the condition Tyrosinaemia type 1 (TYR1) have problems breaking down the amino acid, tyrosine from the food they eat. Left untreated, the condition can lead to the build-up of harmful chemicals which can cause damage to vital parts of the body, such as the liver.

9.1 In 2015, the UK NSC reviewed the evidence to screen for a group of amino acid disorders, and found TYR1 to be a potential screening candidate. It was agreed that for TYR1 to be considered further work needed to look at:

- the epidemiology of the condition in Europe
- studies determining the feasibility of screening for TYR1 in the UK
- the proportion of TYR1 cases currently detected through PKU screening
- the use of succinylacetone (SUAC) as opposed to using tyrosine as a more accurate marker for screening

9.2 The latest review carried out in 2016 found that the areas of concern highlighted remained uncertain. Precise number of babies affected by TYR1 in the UK is unknown. This means that when examining how effective the offer to screen is, the conclusion would be ambiguous as the variation reported currently is significant. As suggested from the previous review, the use of SUAC as the primary marker to detect TYR1 reported good test performance, as being more sensitivity and specific. However the studies examined were found to have a high risk of bias, suggesting that the conclusions drawn from the studies examined should not be taken as wholly reliable.

9.3 Dr Elliman informed the group that a workshop was held in January 2017 as requested by the Committee. It brought together clinical, academic, public health and patient

expertise to discuss in more detail the review findings, to examine the data and suggest whether and what further work is needed.

9.4 The group came to the conclusion that many of the questions would only be answered if screening were to be done. But other screening programmes, outside the UK, may be able to provide the following information: the follow up on the false negative screening results, reanalysis of the outcomes data in the nitisinone treatment and to request for a modelling exercise to be undertaken to estimate the benefit of screening.

9.5 The UK NSC agreed that screening for Tyrosinaemia Type 1 should not be recommended and awaits the outcome of the modelling work to address areas of uncertainty.

The Condition		
1	The condition should be an important health problem as judged by its frequency and/or severity. The epidemiology, incidence, prevalence and natural history of the condition should be understood, including the development from latent to declared disease and/or there should be robust evidence about the association between the risk or disease marker and serious treatable disease	Prevalence of TYR1 in the UK remains unknown
The intervention		
9	There should be an effective intervention for patients identified through screening, with evidence that intervention at a pre-symptomatic phase leads to better outcomes for the screened individual compared with usual care. Evidence relating to wider benefits of screening, for example those relating to family members, should be taken into account where available. However, where there is no prospect of benefit for the individual screened then the screening programme shouldn't be further considered.	It is clear that early treatment for TYR1 is beneficial when compared to later treatment however further work is needed to understand what impact screening has on the false negative screen result babies as well as look at other research using nitisinone
13	The benefit gained by individuals from the screening programme should outweigh any harms for example from overdiagnosis, overtreatment, false positives, false reassurance, uncertain findings and complications.	

Screening for Vasa Praevia in the second trimester

10. This item was presented by Dr Anne Mackie who for the benefit of the Committee described the condition of Vasa Praevia, as being a complication that can occur in pregnancy when the baby's blood vessels cross the cervix. There is an associated condition where the blood vessels run outside of the protective layer. When the cervix dilates or membranes rupture in labour, the unprotected vessels can tear and cause significant bleeding to the mother and baby.

10.1 The first review of vasa praevia was in 2013 which found that there was insufficient evidence when looking at: case definition, natural history, epidemiology for vasa praevia as well as raising concerns around the use of the test and treatment. As this was the first time the UK NSC had considered the evidence for vasa praevia and given the interest and need for further work, the UK NSC held a workshop which led to several key actions. Most notably was the group's support for guidance in high risk groups to be developed by the Royal College of Obstetrician and Gynaecologist (RCOG).

10.2 Whilst the guidance work led by RCOG is in development, the UK NSC undertook its regular review, as per its published evidence review process. Dr Mackie informed the group that the second review, this time round looked at Velamentous cord insertion (VCI) as well as VP as advised by the FMCH group.

10.3 When reviewing the evidence this time round, the incidence of the condition remains uncertain but assumed to be quite rare clinically. It is the treatment option for vasa praevia as being delivery via an early caesarean section, which causes considerable amount of unease. Dr Mackie highlighted that it would need to be absolutely clear in what circumstances you would offer such an invasive treatment. Furthermore the literature to better understand the test was found to be poor, in that it couldn't estimate how many babies could be correctly detected as being screen positive and test positive.

10.4 The Committee agreed that it was clear that further work was needed to better explore the test as well as the relationship of VCI and vasa praevia. The Committee did express interest in the pending publication from the UK Obstetric Surveillance System (UKOSS) which is hoped will address some areas of concern raised by the UK NSC review. With this and the upcoming guidance for high risk women, the care for women identified as having vasa praevia will clearly be benefitted. The UK NSC emphasised that it stands ready to review any new significant peer-reviewed evidence which may alter a current recommendation.

10.5 The UK NSC recommended that whole population screening for vasa praevia is not introduced.

The Condition		
1	The condition should be an important health problem as judged by its frequency and/or severity. The epidemiology, incidence, prevalence and natural history of the condition should be understood, including development from latent to declared disease and/or there should be robust evidence about the	There is not enough information about the incidence of vasa praevia in the UK; although clinically detected it is rare
4	There should be a simple, safe, precise and validated screening test	More knowledge is needed on the accuracy of the test
The Intervention		
9	There should be an effective intervention for patients identified through screening, with evidence that intervention at a pre-symptomatic phase leads to better outcomes for the screened individual compared with usual care. Evidence relating to wider benefits of screening, for example those relating to family members, should be taken into account where available. However, where there is no prospect of benefit for the individual screened then the screening programme shouldn't be further considered.	No established management pathway for VCI

Screening for Severe Combined Immunodeficiency (SCID)

11. Severe combined immunodeficiency (SCID) is a rare, inherited condition which causes abnormalities to the functioning of the immune system resulting in the infant being unable to fight off infections. The condition is also known as the bubble boy/ bubble baby disease. Treatment for this rare condition is via a stem cell transplant.

11.1 The UK NSC reviewed the evidence to screen for SCID in 2013 and recommended that screening should not be implemented. This was because there was insufficient information and lack of evidence to address all of the UK NSC's criteria which included:; epidemiology of the condition in the UK, the test performance and the clinical and cost effectiveness of offering screening compared to current practise. A concern with screening for SCID was that the test was not specific enough to just detect SCID, it would also screen positive for other conditions. The Committee agreed that although screening for SCID demonstrated potential benefits of early diagnosis it was equally concerned with the harm that could arise from screening, causing unnecessary worry to

many families. The UK NSC commissioned ScHARR to undertake a cost effectiveness evaluation to screen for SCID looking at the benefit of early treatment as well as the harms. ScHARR presented their report at the June 2016 UK NSC meeting. The model estimated that screening for SCID had a 65% chance of being cost effective. The Committee were encouraged by the findings but aired concerns over the robustness of some of the model's assumptions as well as the harms caused by screening for the false positives detected. The UK NSC recommended that a technical appraisal of the evaluation model be undertaken whilst an evidence review is undertaken.

11.2 The UK NSC was presented with the appraisal of the model, which was discussed in depth at a workshop, set up to explore some of the assumptions of the model and discuss next steps. The workshop was attended by experts in the field along with patient and public representatives. The Committee noted that the assumptions made by the model were that:

- there would be an increase in detection for SCID cases. Approximately 17 SCID cases in the UK would be identified each year from population screening of which 5-6 would have been detected via current practise
- the main benefit to offering screening is to detect and prevent infection before the condition presents. The model estimates that without screening eight babies die but could be reduced to two should screening be offered. The model however indicated that of the babies found and treated early they would still have the same health outcomes as those currently detected through current cascade testing.
- approximately 260 families would receive false positive results, with confirmation taking up to two weeks
- early transplantation would not significantly alter long term outcomes in babies who were symptomatic and survive
- the offer to screen is estimated to cost £3.2million a year and depending on some assumptions, is cost effective,

11.1 In addition to this the workshop has also explored the following key areas;

- consanguinity- the incidence of SCID was found to be higher in families where the mother and father had a blood link, meaning that they were related. In the UK over 50% of SCID cases are found in such groups.
- vaccinations- families who originate from countries with high tuberculosis(TB) rate are offered the BCG TB vaccination to their baby soon after birth. Babies who are given this vaccination can suffer from severe complications where SCID has not been detected.
- by products- concern over the non-SCID conditions being detected accidently by the test as it is not accurate

11.2 Mr Marshall summarised some of the key findings from the model if screening were to be introduced as;

- of 780835 live births, 310 would be screen test positives
- of the 310 screen test positives:
  - 17 are predicted to be SCID
  - 260 would have a false positive result
  - 7 are likely to be pre terms
  - 26 are likely to be non-SCID detected conditions

11.3 The UK NSC discussed the model and the evidence review document and concluded that although screening has been suggested as being cost effective the exact number of babies born with SCID in the UK remains unknown. So this would mean that screening would only be effective if it was to detect 17 or more babies as anything less would render the programme was being ineffective and causing more harm.

11.4 An ongoing concern highlighted from the last review was the negative impact screening would have on the false positive babies. The Committee conceded that this was a growing concern and further work is needed to better understand what the implication and effect would be on the 260 families receiving a false positive result would be.

11.5 The Chair summarised the findings and the Committee agreed that screening for SCID, as based on the estimates and parameters of the model, is likely to be cost effective. The UK NSC agreed that the only way to test the model and understand the benefit and harms screening would bring would be to carry out an evaluation of screening for SCID. The Committee agreed that such an evaluation would need clear protocols and be discussed with a working group.

11.6 The UK NSC approved the review document for public consultation and agreed that comments should be brought back to the UK NSC at a later date, with a view to consider that a practical evaluation of newborn screening for SCID be undertaken.

Updates

NIHR NETSCC Update (for information)

The Committee noted the updates

SIGN Update (for information)

The Committee noted the updates

AOB

i. Professor Duffy invited members of the UK NSC to attend a symposium held by PRU at QML. Invitation would be circulated via Ms Mauthoor