

# NUFFIELD COUNCIL ON BIOETHICS

## Non-invasive prenatal testing

### Summary of consultation responses

Published June 2017

#### Contents

Introduction .....	3
Question 1 - If this recommendation was implemented fully into NHS antenatal care, what benefits or concerns might this raise for pregnant women and their partners?..	4
Question 2 - What do you think might be the implications of offering NIPT as part of NHS antenatal care for the healthcare professionals involved in providing prenatal screening?.....	17
Question 3 - If this recommendation was implemented fully into NHS antenatal care, it might lead to an increase in the number of terminations of pregnancies with a diagnosis of Down's syndrome, Patau's syndrome or Edwards' syndrome. What benefits or concerns might this raise? .....	23
Question 4 - Do you think the UK National Screening Committee's criteria for appraising the viability, effectiveness and appropriateness of a screening programme are appropriate for appraising prenatal screening programmes? .....	31
Question 5 - How would you rate the information and counselling currently provided by the NHS to pregnant women and their partners to help them make decisions about currently available prenatal screening (e.g. using ultrasound) for genetic conditions during pregnancy, if you have experience or evidence relating to this? ..	38
Question 6 - How would you rate information and/or counselling provided by the NHS about NIPT available as part of research studies or through the private sector, if you have experience or evidence relating to this? .....	42
Question 7 - How would you rate the information and/or counselling currently provided by private healthcare clinics to pregnant women and their partners to help them make decisions about NIPT, if you have experience or evidence relating to this?.....	46

Question 8 - What information about NIPT and the conditions being tested for do you think should be conveyed to pregnant women and their partners? How do you think that information could best be conveyed and by whom? .....	51
Question 9 - What might be the implications for the NHS of increasing numbers of pregnant women purchasing NIPT through the private sector? .....	57
Question 10 - What benefits and concerns might be raised if pregnant women were able to purchase NIPT directly from providers (e.g. where a kit is sent to the pregnant woman in the post), rather than through a healthcare clinic following a face-to-face consultation? .....	61
Question 11 - A small proportion of NIPT tests will return an inconclusive result, even if repeated. How should healthcare professionals, both in the NHS and in private clinics, deal with inconclusive results? .....	66
Question 12 - What issues are raised by incidental findings that can arise following NIPT (such as genetic abnormalities or cancerous cells in the pregnant woman), both in the NHS and in private clinics? .....	69
Question 13 - Should potential parents be able to find out the sex of their unborn baby for non-medical reasons from 10 weeks of pregnancy using NIPT? Please give reasons for your answer .....	74
Question 14 - What genetic information, if any, do you think parents should be allowed to find out about their unborn baby using NIPT? Please give reasons for your answer.....	79
Question 15 - What genetic information, if any, do you think parents should not be allowed to find out about their unborn baby using NIPT? Please give reasons for your answer.....	83
Question 16 - Do you think whole genome sequencing of unborn babies using NIPT should be allowed? Please give reasons for your answer.....	88
Question 17 - What, if anything, might the increasing availability and use of NIPT mean for people living with genetic conditions? Please provide evidence or examples if possible. ....	94
Question 18 - Is current regulation covering the provision and marketing of NIPT in the UK sufficient and appropriate? .....	100
Question 19 - What ethical values do you think are important or relevant in the context of NIPT? .....	107
Question 20 - Any other comments: Please tell us anything else you would like to raise in relation to NIPT .....	115

## Introduction

The Nuffield Council on Bioethics published a consultation document on 19 May 2016 seeking views on issues relating to recent developments in NIPT. The deadline for responses was 1 August 2016. The consultation document comprised 20 questions, all of which asked for free-text responses.

The consultation was promoted using social media and mailing lists, targeting a range of organisations with members or links to people who might have an interest in NIPT.

The survey received 28 responses. A list of respondents is below.<sup>1</sup> This document summarises the responses received to each question.

## Organisations

Anscombe Bioethics Centre  
Association of Genetic Nurses and Counsellors  
BioCentre  
British Maternal and Fetal Medicine Society (BMFMS)<sup>2</sup>  
British Medical Association (BMA)  
British Pregnancy Advisory Service (BPAS)  
Christian Action Research and Education (CARE)  
Christian Medical Fellowship (CMF)  
Church of England, Mission and Public Affairs Council  
Clinical Genetics and Cytogenetics, Guy's Hospital  
Down's Syndrome Association (DSA)  
Down Syndrome Research Foundation UK (DSRF UK)  
Genetic Alliance UK  
Jane Fisher, Director, Antenatal Results and Choices (ARC)  
PHG Foundation  
Royal College of Obstetricians and Gynaecologists (RCOG)  
Saving Down Syndrome  
The Liminal Spaces Project, University of Edinburgh, funded by the Wellcome Trust (The Liminal Spaces Project)  
WeLDNurses  
Victoria Woodham, on behalf of Future of Down's

## Individual

Felicity Boardman, Warwick Medical School  
Anindita Doig  
Matthew Jolly, National Clinical Director for the Maternity Review and Women's Health, Acute Medical Directorate, NHS England  
Colette Lloyd  
Kay Sammon  
Rachel Siden  
Lorna Watson

---

<sup>1</sup> Respondents who agreed for their responses to be quoted in this summary but did not want these quotes to be attributed to them are described as 'anonymous respondents'.

<sup>2</sup> Three members of the BMFMS responded as part of a collated, organisational response, and are individually quoted in this summary as 'BMFMS members'.

**Question 1 - If this recommendation was implemented fully into NHS antenatal care, what benefits or concerns might this raise for pregnant women and their partners?**

**Benefits**

**1. Potential to reduce number of invasive diagnostic procedures**

A number of respondents said that one of the benefits of implementing NIPT into an NHS antenatal care was that fewer women would need to undergo invasive diagnostic procedures, amniocentesis and chorionic villus sampling (CVS) which carry a risk of miscarriage, in order to find out if their fetus was affected by Down's, Edwards' or Patau's syndromes.

The Association of Genetic Nurses and Counsellors said that:

*The main benefits for women and their partners are avoiding invasive testing and therefore avoiding the associated risk of miscarriage...*

Victoria Woodham, on behalf of Future of Down's (Future of Down's) also made the point that NIPT would give women another option before having to undergo an invasive procedure, to get a diagnosis.

*From an individual patient perspective, the availability of NIPT as an intermediary step between combined screening and amniocentesis removes the concern about potential risks of invasive procedure.*

Jane Fisher, Director of Antenatal Results and Choices (ARC) appealed to evidence suggesting that the number of invasive tests would go down if NIPT is made available in the NHS and that this would mean that there would be a resulting drop in the number of procedure related miscarriages, adding that implementation of NIPT within the NHS should be evaluated.

*ARC is supportive of the UK NSC recommendation to implement NIPT into the current fetal anomaly screening programme in a careful, evaluative way. Evidence suggests this will make the programme more effective. This should enable those women who opt to have screening for Down's, Edwards' and Patau's syndromes to benefit from the fact that the testing is less likely to lead to the prospect of an invasive diagnostic procedure. Thus there will be a reduction in procedure-related fetal loss.*

A member of the British Maternal and Fetal Medicine Society (BMFMS) expressed confidence that the number of procedure related miscarriages would indeed go down.

*The number of miscarriages occurring following invasive testing would undoubtedly fall; clearly a good thing.*

The British Pregnancy Advisory Service (BPAS), which advises women and couples on abortion, pointed out that making a decision about undergoing an invasive

diagnostic test can itself be very difficult for women and couples and it would be beneficial if women were faced with these choices less frequently.

*The introduction of a systematic population screening programme, which would provide an offer of a cell free DNA (cfDNA) test, will be enormously beneficial to women grappling with a decision about an invasive test and consequent risk of miscarriage.*

Individual respondent Felicity Boardman echoed this view and added that fewer invasive diagnostic tests would also mean fewer risks to women undergoing the tests.

*Fewer invasive diagnostic tests carried out which could reduce the number of miscarriages and also risks (physical, psychological) to the mother.*

## **2. Enables women and couples to prepare**

It was considered by some respondents to be a benefit of making NIPT available in the NHS that it could enable women and couples to prepare for the birth of a child with a disability. The Christian Medical Fellowship (CMF) said that NIPT could give women more time to get ready, psychologically, as well as provide the chance to learn more about their child's condition.

*A positive NIPT screening result, followed by a diagnostic invasive test that confirms trisomy, would enable parents to prepare for the arrival of a child with special needs. The availability of time for reflection, qualified counselling and support, written online resources and the opportunity to meet with parents of children with trisomy-related disabilities, and the children themselves, would all be helpful in this.*

Future of Down's agreed that the opportunity NIPT would provide women and couples to prepare for the birth of their child would be beneficial.

*For parents wishing to continue with a pregnancy regardless of whether trisomy is confirmed NIPT presents the opportunity to be more certain of a diagnosis of trisomy and to prepare accordingly.*

WeLDNurses, whose consultation response summarised the findings of one of the twitter chats the organisation holds twice weekly, said that:

*Participants acknowledge that NIPT afforded women and families swifter and more reliable information with which to make choices, which would go some way to enable the necessary preparation for the arrival of their baby.*

## **3. Equitable access to NIPT**

Some respondents pointed out that making NIPT available in the NHS would improve on the current situation in which only those with means are able to access NIPT through the private sector. Offering NIPT to those women with high chance results from the combined test would help to make this situation fairer and more equitable. A member of the BMFMS said:

*Although NIPT is currently available privately, the costs are prohibitive for some women so NHS provision would remove the inequity of access.*

The PHG Foundation made the same point arguing that:

*Implementation in the NHS will address the current inequity of access to this safer non-invasive test arising from extensive commercial availability.*

Future of Down's agreed that current arrangements were imperfect and unfair, and said that making NIPT available in the NHS would address this, adding the caveat that appropriate care, support and information about genetic conditions for those undergoing testing would need to be assured.

*The current availability in some regions through selected medical centres to selected women and private practice leads to confusion, misinformation and inequity. An NHS implementation if carried out with due care for the parents embarking on testing and consideration of equity of resources and options available to parents, can with inclusion of appropriate counselling and information begin to address concerns that testing may have detrimental effect on members of society with disability.*

#### **4. Ease of access**

Some respondents pointed out that NIPT was easier to access and undergo than invasive tests, and cited this as an advantage of the test. The PHG Foundation said that NIPT was:

*Quicker, easier, safer identification of those at low-risk.*

Felicity Boardman said that because the test was relatively easier to carry out and undergo than other screening tests this might make it more straightforward to arrange through the NHS, with potential for fewer delays in access.

*Quick access to the test as it requires less skill to carry out (simple blood draw) than an amnio/ CVS so could be performed by non-specialist. This might mean reduced waiting time for an appointment to have it done and could mean that pregnant women and their partners might get an answer fairly quickly as to whether or not they will be offered a diagnostic test.*

#### **5. Earlier timing**

The fact that NIPT can be conducted earlier in pregnancy than invasive tests was seen to be a benefit by some respondents. The PHG Foundation said one of the main positives for women would be:

*...Receiving the results from NIPT at a much earlier stage in pregnancy compared to results from invasive testing.*

Genetic Alliance UK argued that NIPT might enable earlier diagnoses which would mean that women would have more time to consider their options and could lessen anxiety.

*The earlier diagnosis (from 10 weeks gestation rather than 11-14) for chorionic villus sampling and 16-22 for amniocentesis) allows the woman more time to think about what is the most appropriate decision for her.*

The British Medical Association (BMA) also said:

*Earlier identification of Down's Syndrome, Patau's Syndrome or Edwards' Syndrome allows more time for women to make the right decision for them and their family about whether to continue with the pregnancy.*

## **6. Less discomfort**

NIPT involves less discomfort than invasive diagnostic testing and this was pointed out by the PHG Foundation who observed that many women who receive a low chance NIPT result would be able to avoid the discomfort associated with invasive testing.

*Improved patient satisfaction due to reduction in the decision-making, anxiety and discomfort associated with invasive tests.*

## **Concerns**

### **7. Need for quality information**

Many of the points made in support of the introduction of NIPT to NHS antenatal care were made alongside the caveat that information about NIPT, the conditions it is used to test for and sources of support and services for parents of disabled children, would need to be of a high standard to ensure that women are in a position to give informed consent to the test. The BMA for example said:

*We recognise that concerns about routinisation have been raised previously in relation to prenatal testing more generally, our guidance is that health professionals have a general ethical and legal duty to ensure that patients are given sufficient information to understand what is proposed and are given the opportunity to give or withhold consent. In Medical Ethics Today we advise that "...parents should be given as much information as necessary to enable them to make an informed decision about whether to opt for testing and, if so, how to respond to an unfavourable result" and that "when giving information to patients, health professionals should also present the possibility of refusing all prenatal screening as a reasonable and acceptable option."*

The Down's Syndrome Association (DSA) however raised concerns about expertise and capacity within NHS prenatal screening services to provide women and couples with adequate, up to date information about Down's syndrome. Citing evidence gathered through its own training programmes and helpline, as well as the conclusions of the Royal College of Obstetricians and Gynaecologists report [Non-invasive prenatal testing for chromosomal abnormality using maternal plasma DNA: scientific impact paper no.15](#), they criticised the existing screening programme and raised concerns regarding what this might suggest about the implementation of NIPT on the NHS.

*Based upon the information the DSA has received from its 'Tell it Right, Start it Right' (TIR) training and helpline calls, it is questionable whether the NHS has the capacity to provide the counselling and information necessary for these parents. Scientific Impact Paper No 15 from the Royal College of Obstetricians and Gynaecologists stated that one of the consequences of the*

*implementation of NIPT for the existing services would be that “resources for the education and training of health professionals offering this testing and pre-test information and discussion with the patient will be required”.*

The Down Syndrome Research Foundation UK (DSRF UK) made a similar point about expertise within the NHS and current provision of information on Down's syndrome, suggesting that NHS healthcare professionals are not always aware of the limitations in their understanding of the condition.

*Currently in the majority of clinics there are insufficient skills or time to perform full informed consent into the prenatal screening process. It has been demonstrated there is a lacking within the current delivery of informed consent with a bias for not giving information (professionals) and patients not asking. People and professionals believe they 'know' and are informed about DS but feedback and research repeatedly shows us they are not.*

Individual respondent Colette Lloyd agreed that current arrangements were inadequate and that the situation could be worsened in the event that NIPT is incorporated into the pathway without appropriate preparation.

*Lack of good information about the conditions, the test itself and obtaining truly informed consent is a problem with the current system. The new blood test is only going to exacerbate this issue, without proper precautions firmly in place.*

A member of the BMFMS agreed that information would be vital, stressing that women's needs both before and after testing should be met.

*Offering NIPT on the NHS for those women at high risk raises some concerns: the NHS must provide pregnant women with access to timely and comprehensive information and counselling, both pre- and post-test, including a clear explanation of the options available if the test is positive, to enable women to make a truly informed choice.*

The Church of England, Mission and Public Affairs Council also stressed the need for quality information and counselling and expressed concerns that, without this, the number of terminations of fetuses with Down's syndrome could rise.

*...While recognising that NIPT will provide women with greater information during their pregnancies, as stated above, we are concerned that an unintended consequence of this might be an increase in terminations of pregnancy. Unless NIPT is accompanied by comprehensive, accessible information with any subsequent diagnostic tests being accompanied by adequate counselling, there is a real possibility that NIPT could become a catalyst for further abortions on the grounds of foetal disability.*

Felicity Boardman concurred, adding that people living with the conditions for which NIPT tests should be involved in developing the information that women and couples considering prenatal screening receive.

*The education and information surrounding NIPT would need to be high quality, and delivered in consultation with families living directly with the conditions.*

Both Christian Action Research and Education (CARE) and BioCentre cited the conclusions of the 2013 [Parliamentary inquiry into abortion on the grounds of disability](#) which found that, amongst other things, information made available to women and couples about sources of support for parents of disabled children was lacking.

*The Inquiry found... there was a need for information about support services that can help parents who choose not to abort their disabled child; including adoption and palliative care options.*

The Liminal Spaces Project, University of Edinburgh (The Liminal Spaces Project) said that information about available support would be especially crucial in the event that an NIPT result is high chance, and involving a range of care professionals in discussions would be important. They acknowledged, however, the challenges involved in providing this quality of service for women.

*Parents ought then to be supported to make decisions about how to proceed, and this has consequences that once again transcend simplistic divides between, say, private testing and public services. For example, social workers could be included in genetic counselling sessions in order to share information regarding support services available for patients living with a disabled child. We are not so naïve as to imagine, however, that such services will always be available. This emphasises all the more the importance of a robust informed consent process.*

They also cited the importance of confronting all relevant issues in ensuring that women are in a position to give their informed consent to undergo NIPT.

*Informed consent should include explicit conversations about issues related to abortion, disability, and whether women really want the information provided by NIPT, as well as explanations of the tests available, how they work, their limitations, and the conditions that can be detected.*

Some respondents were concerned about women and couples' understanding of the meaning of an NIPT result, and the need for further diagnostic testing, to confirm NIPT results. The Church of England, Mission and Public Affairs Council highlighted the need to ensure that the fact that an NIPT result does not constitute a diagnosis was conveyed.

*It is essential that women are given accurate information with regard to the distinction between screening and diagnosis as well as to the possible implications of screening results. This must include information on the reliability of NIPT screening and the need for subsequent diagnostic tests (and their risks) in the event of screening indicating a high level of probability that a chromosomal abnormality is present.*

The CMF suggested that there is currently misunderstanding in this area and cited the findings of one US study indicating that women do not always understand, following discussions with healthcare professionals, that screening tests are not diagnostic.

*The public misconception that NIPT is a 'diagnostic' test, on a par with invasive testing, not a screening test that would need to be followed by a diagnostic invasive procedure. Research has found that in over half of discussions health care providers did not clarify the fact that screening is diagnostic.*

It was pointed out by the CMF that once implemented into NHS antenatal care, awareness of NIPT amongst the public would be likely to increase, which may result in more women with a lower chance of having an affected pregnancy seeking the test privately. Quality of information and support in the private sector was a key concern for the CMF.

*As awareness of the test increases, and its cost comes down, then many pregnant women will seek to access the test privately. They may not receive pre-test information and counselling. They will receive results outlining all manner of variable predictive risks faced by their babies, but will not have the context in which to discuss, assess and weigh the relevance of those results. This will increase anxiety further and make abortion a more likely outcome, sometimes without evidence of trisomy.*

## **8. Potential for delays in diagnosis**

Future of Down's pointed out that there may be delays for women in undergoing invasive diagnostic procedures once NIPT is made available on the NHS, arguing that these delays may result in increased anxiety.

*...Where the parents are considering termination if trisomy is identified, NIPT testing may introduce a delay between receiving combined results and conduct of a confirmatory invasive diagnostic test that would not be present if NIPT is not undertaken. This delay may have the potential to increase anxiety in parents and lead to what they may consider as an unacceptable delay in accessing the desired termination. It will be important to ensure that parents embarking on this route are aware of this and choose the appropriate testing pathway for their situation.*

ARC made the same point, emphasising that any delays in obtaining a diagnosis may limit a woman's termination options.

*...it does add an extra step into the screening pathway which may delay diagnosis for some. This could be an issue for those women who have trisomy diagnosed and decide to have a termination. If their diagnosis is made by amniocentesis it is likely that their NHS provider will not be able to offer a choice of termination method.*

The problem might be exacerbated, ARC argued, by the fact that there are already issues with the availability of surgical terminations on the NHS.

*There is currently a serious lack of provision of surgical termination beyond 13 weeks in NHS hospital settings.*

Felicity Boardman stressed that in addition to restricted options for termination at a later stage, some women would find the experience of terminating a pregnancy further into gestation a more difficult experience.

*This has implications for the woman (in terms of emotional stress), but could also negatively impact on her decision-making around termination of pregnancy, or render termination of pregnancy more distressing (due to advanced foetal age). In short, NIPT could leave these women 'in limbo' for longer, as both the screening test and NIPT are not diagnostic.*

BPAS agreed, arguing that the NHS should address this issue by partnering with external providers of surgical abortions.

*The NHS must acknowledge that an extra layer of screening will delay diagnosis. It is inevitable that women who opt for cfDNA screening and go on to need an invasive diagnostic test will be making a decision about their pregnancy at later gestations than if they had one test. For the majority of women with a positive diagnosis who choose to end their pregnancy this will mean it may be harder to obtain a choice of method of termination. While some women will request a medical termination others will find the prospect very distressing and would prefer a surgical termination. As the NHS is unable to provide surgical abortions at later gestations in many areas it must ensure that it continues to develop its relationships with independent third sector providers. The need for surgical abortion in the second trimester is likely to grow as a result of the additional screening test and the NHS must ensure that services are able to meet women's needs post-diagnosis.*

## **9. Incidental findings and inconclusive results**

Some concerns raised by respondents related to inconclusive results and incidental findings about the health of the woman undergoing the test. Clinical Genetics and Cytogenetics, Guy's Hospital raised "*incidental findings/sex chromosome anomalies*" as a concern.

The BMA were concerned with the issue of how NHS doctors should manage incidental findings which, it said, "*have the potential to raise significant issues for healthcare professionals.*" Challenges related to informing women about these findings and risks of the potential for over-diagnosis and unnecessary treatment were raised. A better understanding of this area was needed, the BMA said.

*...It raises questions about whether the information should be disclosed when it may or may not be clinically significant and when it could lead, in some cases to overdiagnosis and unnecessary investigations and treatment. In our view, more research is needed to inform discussion and stakeholder guidance to health professionals on this issue.*

The BMA cited its advice in [Medical Ethics Today](#) which could be relevant for healthcare professionals dealing with incidental findings associated with NIPT.

*If there is a reasonable chance of other information being inadvertently discovered from a particular test, this should be discussed with the patient...during the consent process in order to ascertain the individual's wishes about disclosure. The discussion should give examples of the type of information that could be discovered and procedures that will be followed in that event.*

Inconclusive results were raised by Clinical Genetics and Cytogenetics, Guy's Hospital who argued that these, and the delays to which they give rise, could increase anxiety.

*Inconclusive/failed results will prolong testing pathway and may result in greater anxiety and more invasive tests.*

Future of Down's response claimed that the proportion of women who will receive inconclusive results was relatively high and that it was important, therefore, when seeking consent to discuss this possibility with women and couples.

*NIPT is commonly (between 1-10%) inconclusive; parents must be informed of the potential need for retesting and the possibility that no definitive result may be achieved as a result of testing.*

## **10. Financial costs**

The issue of how introducing NIPT as part of an NHS antenatal care would impact on NHS budgets was raised by some respondents. Colette Lloyd described NIPT as "...a huge expense that the NHS can hardly afford..." and a member of the BMFMS raised a question about the opportunity costs associated with introducing NIPT into the NHS prenatal screening pathway.

*There will of course be cost implications. If this is not cost neutral, what other area of maternity care will suffer to fund this test?*

Clinical Genetics and Cytogenetics, Guy's Hospital made the point more strongly.

*...Increased cost to the NHS, draining resources from other needs.*

On the other hand, Genetic Alliance UK argued that making NIPT available to women in the NHS would be unlikely to result in further costs, citing economic assessments conducted by the UK National Screening Committee.

*Detailed health economic analyses carried out by the UK NSC suggest that implementing NIPT as recommended would result in little extra cost to the NHS, or may in fact be cost neutral.*

## **11. Impact on people with genetic conditions**

Another concern raised by some respondents related to the impacts on people with the conditions for which NIPT is used to test. One concern was that there may be fewer babies born with Down's, Edwards' and Patau's syndromes as a result of making NIPT available in the NHS and that people with these conditions would be

effectively 'screened out'. CARE argued that this may be one consequence of widening availability of NIPT describing the impact as potentially 'eugenic'.

*CARE is of the view that NIPT would negatively impact babies with trisomies, such as Down's syndrome – effectively having an inadvertent eugenic effect – essentially screening them out.*

BioCentre took precisely the same view.

*NIPT would negatively impact babies with trisomies, such as Down's syndrome – essentially screening them out.*

DSRF UK went further, arguing that NIPT would worsen discrimination faced by people with genetic conditions and disabilities, and their families.

*We believe NIPT will not only perpetuate discrimination against people born and unborn with DS (Downs syndrome), but also their parents.*

Future of Down's was also concerned about this but indicated that this risk could be mitigated by ensuring that counselling, information and support for women and couples was adequate.

*An NHS implementation if carried out with due care for the parents embarking on testing and consideration of equity of resources and options available to parents, can with inclusion of appropriate counselling and information begin to address concerns that testing may have detrimental effect on members of society with disability.*

## **12. Risks associated with the involvement of the private sector**

A number of issues related to the involvement of the private sector were raised by respondents. It was suggested that private and commercial interests might be driving the use of NIPT in ways that do not necessarily promote public health.

The DSA pointed out that making NIPT available in the NHS only to 'higher chance' women (those identified as having a greater than 1 in 150 chance of having a fetus after the combined test) might mean other women would seek NIPT in the private sector. This might be problematic due to the quality of information and counselling made available by private clinics offering NIPT, DSA said.

*The intention is to implement NIPT only for the high chance group, which means the test will continue to be available privately to other women. This is of concern as there is not yet a clear picture of the quality of information provided prior to testing in the private sector.*

The CMF made a similar point, observing that growing public awareness of NIPT which would be likely to increase further following the integration of NIPT into NHS antenatal care, raised issues insofar as more women would be likely to pursue NIPT in private settings. Private providers, they said, may be less likely to support women and couples and might make available to them information that could be difficult to fully understand (see p.10).

Colette Lloyd highlighted the ways that antenatal care might be complicated by the introduction of NIPT to NHS antenatal care in combination with the ongoing availability of NIPT in the private sector. She pointed out that women who receive a low chance result from the combined test, but a high chance result from a privately accessed NIPT, may want to undergo invasive diagnostic testing. This, she said, might counter the reduction in invasive tests.

*It may reduce the number of amnios based on the RAPID study but in actuality the new test may even increase number of amnios as there will be women coming from the private sector having been given an inaccurate high risk NIPT and therefore asking for an amnio, when in the past they wouldn't have been offered one from the combined screen.*

### **13. Integration into NHS antenatal care 'endorses' test**

Some respondents said that by introducing NIPT to NHS antenatal care, a message about Down's, Edwards' and Patau's syndromes was being conveyed to women and couples, and the wider public. Some were concerned that this might be seen by women and couples as an indication that the state thought they *should* undergo prenatal testing. The CMF said that making NIPT a part of NHS antenatal care might amount to a:

*...Reinforcement of the notion that children with chromosomal abnormalities (and thereby special needs) should be 'screened out' and destroyed.*

The DSA raised a similar issue suggesting that making the test part of NHS antenatal care would legitimise use of the test for Down's, Edwards' and Patau's syndromes and create a risk that women and couples may assume, consciously or otherwise, that they should make use of it.

*The DSA is concerned that pregnant women may see NHS ratification of NIPT as a badge of approval and take the test without considering whether they really want it.*

Felicity Boardman agreed that women and couples, observing that considerable resources have been directed towards making NIPT for these conditions available, may draw conclusions about the harms of having, or parenting children with, these conditions.

*By investing in further tests in the screening process, the fact that the conditions screened-for are important to avoid is underscored. Pregnant women and their partners may feel that they should avoid these conditions because such lengths have been taken to ensure that they can have the option of selective termination of the pregnancy.*

### **14. Pressure to test or to terminate**

A related point concerned pressure to undergo NIPT or to terminate an affected pregnancy. The Church of England, Mission and Public Affairs Council said:

*It is essential that women are fully informed of all potential implications of NIPT and offered it without pressure to agree to the procedure.*

It was said by the Association of Genetic Nurses and Counsellors that the ease of NIPT might contribute to this pressure.

*The main concerns are that pregnant women and their partners may feel pressured into having this test if it is part of routine care. AGNC members are aware of anecdotal reports that this is happening with amniocentesis for pregnancies found to be at high risk on serum screening, but this may be even more likely as it is 'just a blood test' and they will be having regular blood tests as part of antenatal care.*

BioCentre and CARE raised concerns about assumptions that might be made by members of the medical professions, which were raised in the 2013 [Parliamentary inquiry into abortion on the grounds of disability](#):

*It was perceived that medical professionals assumed that parents ought to opt for abortion if their child was diagnosed with having a fetal disability*

A related concern raised by the CMF was that the availability of NIPT might result in more women experiencing 'tentativeness' in pregnancy and finding it hard to commit to their pregnancy until prenatal testing had been carried out. This might mean, in some cases:

*...The mother is wary of committing emotionally or relationally to her unborn child lest the baby be shown by tests to carry an abnormality*

## **15. Grief associated with termination**

Some respondents made points about the psychological impacts of abortion on women and suggested that this should be taken into account, if it were possible that making NIPT available in the NHS could give rise to an increase in terminations. The CMF said that women could experience mental health problems after undergoing terminations and cited the 2013 [Parliamentary inquiry into abortion on the grounds of disability](#) in support of this claim.

*Increased likelihood of some women making choices they later regret. A British Parliamentary Inquiry into abortion on the grounds of disability concluded that: '...the studies have all found that around 20% of women, between one and two years after an abortion for fetal abnormality, have a psychiatric condition, usually a complicated grief reaction, a depressive disorder or post-traumatic stress disorder.' The availability of perinatal palliative care would encourage a higher proportion of pregnant women carrying a baby with a trisomy disorder to continue their pregnancies and avoid many of the mental disorders associated with regret.*

BioCentre cited evidence which they argued had found that psychological impact of terminating a pregnancy at an advanced stage can be similar to that of experiencing a miscarriage.

*Following termination there is a significant risk of the woman experiencing mental health complications. The grief women experience as a result of*

*terminating pregnancies at advanced gestation for fetal anomalies can be as intense as those who experience spontaneous perinatal loss, with approximately a fifth developing major depression and/or requiring psychiatric intervention.*

## **16. Differences between screened-for conditions**

The Church of England, Mission and Public Affairs Council said that differences in the ethical issues raised by screening for Patau's and Edwards', and Down's syndrome should be acknowledged.

*We recognise that Patau's syndrome and Edwards' Syndrome are of such severity that many prospective mothers might feel unable to continue with their pregnancies in view of the potential suffering of any children born and the emotional trauma involved in caring for them. Down Syndrome, however, is quite different, with very many individuals living happy and fulfilled lives, giving great joy to their families and contributing richly to the diversity of society.*

## Question 2 - What do you think might be the implications of offering NIPT as part of NHS antenatal care for the healthcare professionals involved in providing prenatal screening?

### 1. Specialty healthcare expertise

Respondents made a range of points about the consequences for healthcare professionals of making NIPT available on the NHS. Some respondents said the expertise of particular kinds of healthcare professional was crucial for a successful implementation of NIPT on the NHS. A number made comments about the need for particular kinds of healthcare professional, suggesting in some cases that recruitment of more staff would be necessary.

One field of expertise that was seen as critical by some was genetic counselling. Colette Lloyd cited the National Society of Genetic Counsellors' recommendation that all women undergoing NIPT should receive genetic counselling.

*...The National Society of Genetic Counsellors have strongly advised that any woman receiving this test receives counselling from a genetic counsellor before undergoing testing. I would agree that this is needed, in fact this would be a good idea anyway, even under the current regime of testing.*

Genetic Alliance UK pointed out that the RAPID findings had suggested that "...patients have shown a preference for receiving pre and post test counselling from a specialist genetic counsellor" and Future of Down's also stated that genetic counsellors should be involved.

*Future of Down's considers Genetics Counsellors to be most appropriately qualified to attend parents in decision making process regarding NIPT... Future of Down's notes there is a profound shortage of experienced Genetics Counsellors in the UK.*

Questions about whether there was large enough stock of trained genetic counsellors to fulfil this role were also raised by the DSA.

*Are there sufficient numbers of genetic counsellors available to support women in the choices they make?*

DSRF UK suggested that "new professionals may need to be added to the team to assess and weigh up the information NIPT is bringing to each pregnancy" and posed a question over who would have primary responsibility for discussing screening with women.

*Who is the gatekeeper of the screening pathway? Currently this is often delegated to a midwife or sonographer who is unlikely to have sought or had extra training on the conditions she is discussing, beyond a list of co-morbidities. This will be for a variety of reasons, time, unaware of necessity, not mandatory or available.*

WeLDNurses response suggested that there was a role for learning disability nurses in the screening process by, for example “...working with maternity services to plan the ‘screening’ pathways to identify points at which adjustments could be made” or working to “...provide and facilitate education and training opportunities to frontline maternity staff, relating to learning disabilities and reasonable adjustments....”

*It could be expected that these collaborative activities would have positive implications for women and families going through these screening processes while also providing support to midwives in supporting women with learning disabilities and learning difficulties using maternity service.*

## **2. Need for training and education of all involved healthcare professionals**

A number of respondents cited the need for training and education within the healthcare profession to accommodate NIPT within NHS antenatal care. Within this point, respondents said that high quality counselling, a good understanding of the technical aspects of NIPT and knowledge of the conditions for which NIPT is used to test were important.

Technical skills in analysing samples were seen to be important by the PHG Foundation who said that this might have implications for staffing and training.

*Depending on the mode of provision, laboratory and technical staff would also need to be trained and recruited to provide the testing service, including the capacity to collate data from these tests, evaluate their effectiveness and integrate that knowledge into existing and future practice.*

A member of the BMFMS said an appreciation of the performance of the test and its accuracy for different conditions would be necessary.

*Healthcare providers will need to understand clearly how the test works. Specifically, they will need to know its limitations and its positive predictive value for different aneuploidies in different settings.*

A number of respondents stressed the importance of ensuring that healthcare professionals involved in the delivery of NIPT had a good, up to date understanding of chromosomal disorders and were able to relay accurate, balanced information about the conditions. The CMF said:

*CMF supports the recommendations of the Parliamentary Inquiry into Abortion on the Grounds of Disability (2013) that include: ‘Guidelines for the medical professional should include training for obstetricians, fetal medicine specialists and midwives on the practical realities of the lives of children living with the different conditions which are screened for through ante-natal tests.*

It was suggested by Future of Down’s that the need for this kind of training was urgent.

*There is an immediate need for training of all healthcare professionals involved in prenatal screening. This need is absolute and is already evidenced by inadequate responses to public concerns which exist as a result of misinformation prevalent in the media and by misrepresentations of NIPT in the private sector. Healthcare professionals are insufficiently informed and are*

*providing inaccurate advice to women about the conduct of NIPT even in their own centres.*

Colette Lloyd concurred that this area of training is currently not adequate in the UK.

*Healthcare professionals will need to update their training and their understanding of the conditions being tested for. ...we have yet to develop good support for doctors supporting women in this country.*

It was said that specialist organisations should be utilised to support the training of healthcare professionals and development of information. The DSA said:

*The NHS should work more closely with specialist organisations like the DSA who are able to provide information and training to support the work of healthcare professionals involved in the delivery of the screening programme.*

WeLDNurses said that participants to their twitter chat on NIPT thought that involving people with personal experience in training was important.

*Participants were enthusiastic to identify creative, engaging, and experiential education opportunities to support and enable frontline staff to reflect on their values and beliefs while identifying adjustments and improvements to the care and support that all women and families might experience. It was observed that personal testimony was the most powerful, and involving people with learning disabilities, family members and other stakeholders would be the recommended for local healthcare providers and higher education providers.*

Genetic Alliance UK observed that given there exists a “mismatch” between the availability of genetic counsellors of whom they said there were only “300 individuals listed on the GCRB’s Register of Genetic Counsellors” and the need that would be created by making NIPT available to women on the NHS, it was imperative that midwives, obstetricians and other healthcare professionals who would be likely to discuss NIPT with women and couples were adequately trained.

*This lack of specialist knowledge makes it all the more necessary that generalist health care professionals are suitably trained to provide this service.*

Colette Lloyd said that healthcare professionals should be trained in discussing the possibility of adoption with women.

*Healthcare professionals must also be trained in the third option in pregnancy, that of having the baby then giving it up for adoption. Great Western Hospitals and Royal Devon and Exeter Hospitals do mention this in their booklets, however it is notably absent from many other booklets. It is essential that healthcare professionals are trained to convey to women that there are three options, termination, continuation or continuation and then adoption.*

It was added that it would be important to ensure that healthcare training and standards throughout the country were uniform. The DSA queried:

*Will there be equity in the quality of information available across the country?*

### 3. Impact on healthcare professionals delivering invasive procedures

Some responding to this question raised potential issues surrounding maintenance of skills required for invasive procedures. Clinical Genetics and Cytogenetics, Guy's Hospital said:

*Reduced number of invasive tests may de-skill clinical and laboratory staff and increase costs of laboratory tests.*

A member of the BMFMS suggested that training and services would need to be centralised in order for these skills and standards to be maintained.

*Widespread use of NIPT may also have significant implications for some existing services. Invasive prenatal testing methods require a great deal of skill and experience from the health care professional performing the procedure, and there is a known relationship between the skill of the person carrying out the test and the rate of procedure-related miscarriage ([Amniocentesis and Chorionic Villus Sampling \(Green-top Guidelines No. 8 \(2010\) Royal College of Obstetricians and Gynaecologists\)](#)). It will be important to ensure that with the expected reduction in invasive testing as a result of implementation of NIPT, each health care professional is carrying out sufficient tests each year to retain his or her competency.*

The negative potential implications for patients of this outcome was also pointed out by the PHG Foundation.

*The absolute number of invasive tests is expected to fall substantially, which could in turn lead to an overall reduction in skilled practitioners, and invasive testing only being offered in highly specialist centres.*

Though it was noted that centralising services might bring benefits too.

*Decreased numbers of invasive procedures resulting in many units withdrawing that part of the service because of insufficient numbers to maintain skills. This in turn would lead to increasing centralisation which may be perceived as a good thing by some but will cause geographical issues for others.*

### 4. Time, resource and workload implications

A range of related points were made about availability of adequate NHS resources, including issues relating to time pressure, expertise and workload for healthcare professionals, given new requirements that making NIPT available to women with a high chance of an affected pregnancy in the NHS would bring. It was pointed out by a member of the BMFMS that uptake of screening may increase.

*I think it is likely that there will be a rise in uptake of screening simply to be pre-warned of the possibility which may affect cost calculations.*

The DSA posed a question about NHS staffing and capacity issues.

*Does the NHS have the capacity to provide the numbers of healthcare professionals necessary to deliver NIPT who are trained in the delivery of*

*balanced, up to date information about Down's syndrome to pregnant women?*

Some pointed out that making NIPT available to women as a second tier test would place more demands on healthcare professionals' time. Colette Lloyd said:

*There will be increased pressure on the time of the healthcare professional to ensure they meet this legal requirement and obtain proper informed consent for the testing.*

Some expressed concern that there were currently insufficient resources, in terms of time and skillsets, within the existing screening programme, for this to happen. Future of Down's said that this had been overlooked by the UKNSC.

*Factors for consideration include who will counsel parents with high chance results on combined screening of the implications for embarking on testing with NIPT. Published data show concern about the quality of informed consent for parents choosing to undergo NIPT (Van den Heuvel et al 2010), However, the UK NSC evaluation to include NIPT as part of NHS screening did not include consideration to supplying any additional resources to address this concern (e.g. access to appropriately qualified counsellors, or increases in midwife resource and appropriate upskilling) (UK NSC Systemic Review Final Report 2015).*

For Genetic Alliance UK these issues were also a concern.

*A key issue for health professionals of implementing the UK NSC recommendation is likely to be relating to the need for appropriate information provision and genetic counselling support for women and their partners. The rapid implementation and evolution of NIPT raises serious concerns about the capacity of the NHS to provide women with appropriate support in accessing this service, both in terms of availability of appropriate staff, and time commitment.*

## **5. Conscientious objection**

Some pointed out that healthcare professionals might feel conflicted about the ethical issues raised by screening and that integrating NIPT into the screening programme may intensify this. It was observed that if the rate of terminations of affected pregnancies were to increase and/or the number of live births to decrease this might mean that healthcare professionals might make, or want to make, a conscientious objection to NIPT. The CMF said that:

*Healthcare professionals would be enabling a kind of informal eugenics, which would raise issues of conscience for many.*

Saving Down Syndrome said that healthcare professionals had no option but to participate in the screening programme and that some were unhappy being involved for ethical reasons. NIPT, they said, may worsen a situation in which healthcare professionals were expected to deliver services they did not agree should be provided.

*Prenatal screening has been imposed on many professionals who do not hold with its principles, introducing NIPT may simply increase the problems that the imposition brings.*

DSRF UK agreed that healthcare professionals may feel that they are being involved in practices with which they do not agree.

*Implications for HCPs include playing a part in the perpetuation of bias against understanding of DS.*

## **6. Legal issues**

The CMF said that in view of the fact that the Abortion Act did not specify the meaning of the expression 'serious handicap', judgements over what conditions satisfied this description were effectively left to healthcare professionals. This might create difficulties, it was said, and raise concerns for women and couples.

*Ground E of the Abortion Act does not specify what amounts to 'serious handicap.' In practice it is left to individual clinicians to 'interpret' the results of screening tests to parents and to inform them of likely implications. There is potential for significant differences (and even conflict) between doctors as to which disabilities fall within the scope of the law and which do not. This is concerning for parents, practitioners, lawmakers and those with disabilities.*

**Question 3 - If this recommendation was implemented fully into NHS antenatal care, it might lead to an increase in the number of terminations of pregnancies with a diagnosis of Down's syndrome, Patau's syndrome or Edwards' syndrome. What benefits or concerns might this raise?**

### ***Benefits***

#### **1. Choice for women and couples**

Some felt that an increase in terminations would constitute a benefit insofar as it was a manifestation of individual women and couples exercising informed choice in pregnancy. A member of the BMFMS said:

*I believe in informed choice, so I am not concerned per se with this possible outcome...personally I do not perceive this as an issue providing patients are appropriately counselled empowering personal choice.*

However, the same BMFMS member said that pressure to undergo testing should not be applied to patients, also pointing out that some women would be likely to be faced with difficult decisions about continuing a pregnancy that may have ended through natural attrition.

*However, society must avoid reaching a position where terminating for abnormality is 'expected' or 'the norm'. It is the case that now we are formally screening for T13 and T18, we will be posing a few more women with the option of termination when their pregnancy would have come to an end spontaneously anyway.*

Felicity Boardman noted that the reproductive choices which might give rise to more terminations might benefit women, particularly, who tend to have a higher proportion of caring responsibilities than men for children with extra needs.

*It is noteworthy that the research literature suggests that the majority of care work for disabled children is carried out by women, who are also more likely to give up paid employment out of the home to care for their disabled child. Giving pregnant women and their partners the option to circumvent this situation can be regarded as increasing their reproductive autonomy.*

It was pointed out by one anonymous respondent that the experience of giving birth to a child with Down's, Edwards' or Patau's syndrome, having received a low chance screening result, could be very difficult for women and couples. If terminations were to rise because women were making decisions on the basis of more accurate screening results this outcome should be welcomed, it was said.

*Any increase in the termination rate would be due to better detection. Having a baby with a major trisomy following a false negative screen is a traumatic experience for the majority of couples when this occurs. Reducing the frequency of false negative screening results would be a benefit.*

The Association of Genetic Nurses and Counsellors said that these potential benefits would accrue to families too.

*Benefits of this may include reducing the distress to families associated with having a child with such conditions.*

The PHG Foundation agreed and added that any rise might indicate that women who previously would not have terminated their pregnancies may have wanted to pursue further prenatal testing, but might have been deterred by the associated risks of invasive testing.

*The increase in diagnoses may include women who previously sought reproductive choice, but found invasive testing unacceptable as a first line test: NIPT may remove this barrier to testing for some women...*

## **2. Costs of care**

The financial costs of care for those born with genetic conditions was cited as a potential benefit of an increased number of terminations. The Association of Genetic Nurses and Counsellors listed this as one possibility amongst a number of considerations:

*Benefits to this may include...reducing the healthcare/social care costs of treating/looking after patients with such conditions.*

The Anscombe Bioethics Centre also mentioned potential implications for health budgets, but made clear that they thought these were of little overall significance.

*There might be financial gains – just as there might with other cases of life termination (for example, non-voluntary postnatal euthanasia). Such gains are however not worth mentioning in comparison with the severity of the harm, both for the child and the parents, of deliberate termination of some unborn human lives.*

## **Concerns**

### **3. Impact on people with screened-for genetic conditions**

A number of respondents were concerned about how people living with genetic conditions might be affected by an increase in the number of terminations of pregnancies affected by those conditions.

Some did not believe that there would be any benefits. CARE for example, said:

*CARE does not believe that there are any benefits to an increase in the number of terminations of pregnancies with a diagnosis of Down's syndrome, Patau's syndrome or Edwards' syndrome...*

BioCentre expressed concerns about the effects on people with Down's syndrome of an increase terminations of pregnancies diagnosed with Down's syndrome suggesting that "...this would have a long term impact on the Down's syndrome population" and that any effort to eliminate genetic conditions using screening technologies would be:

*Offensive and upsetting to both people with genetic conditions and their families....Such actions undermine the value of their lives and the value that these babies can bring their families throughout the duration of their lives.*

BioCentre, CARE and CMF observed that levels of wellbeing amongst people with some genetic conditions can be very high. CARE said:

*Genetic conditions do not necessarily impede individuals from living valuable and fulfilling lives. A study conducted by Skotko et al 'self-perceptions from people with Down Syndrome' of 284 people with Down's syndrome showed that 99% of people with Down's Syndrome were happy with their lives and 97% liked who they are.*

The CMF also argued that though Patau's and Edwards' syndromes were usually more serious conditions than Down's syndrome, it would not necessarily be beneficial for pregnancies affected by these conditions to be terminated.

*[Children] with Edwards' and Patau's Syndromes may also live for days, weeks, months or even years; even the briefest lives afford opportunities for parental bonding, and support from palliative care and perinatal hospice teams can transform the experience for families.*

CARE also suggested that a rise in terminations might devalue the lives of disabled people and raised a concern about the broader implications for how disability is seen by society.

*Any introduction of screening techniques with the intention of impacting certain types of people, such as babies with genetic conditions, raises significant ethical concerns – particularly about the way that disability is viewed.*

WeLDNurses reported that those who had taken part in their NIPT twitter chat were saddened by the idea that terminations might increase, and suggested that the prospect of this happening highlighted the ways in which their views of disability might differ from those held by other healthcare professionals.

*A question about participants' feelings toward an increase to terminations as a result of the introduction of NIPT elicited an overwhelming response of sadness and provided participants with an opportunity to reflect their own values and beliefs as individuals and how these might align or contrast with those of a registered healthcare professional.*

Future of Downs warned of the potential for increasing numbers of terminations to impact on the birth rate of babies Down's syndrome and challenged claims made by RAPID study researchers that the birth rate would be unlikely to be affected.

*Whilst the rate of terminations may remain at 90% of diagnoses, these are new diagnoses that would not have been made without this technology. Therefore, the net effect is a reduction in live births of individuals with Down's syndrome.*

If there were a lower number of people with Down's syndrome, Future of Down's argued, this could create issues concerning the wider societal understanding of disability, service provision, and societal inclusion.

*Reductions in the population of individuals with Down's syndrome reduces the opportunity to demonstrate the inaccuracy of...prejudices and threatens to further embed these attitudes within society. The reduction of individuals with Down's syndrome has the potential to increase the intolerance in society for those with the condition...and negatively impact quality and availability of provision of resources for early intervention with the downstream effect of reducing opportunity for individuals with Down's syndrome.*

WeLDNurses had a similar concern:

*There were observations relating to the particular subcultures or communities of people with Down's syndrome and their families and supporters, with the implication that these would be significantly impacted by the reduction in the population and the associated isolation, exclusion and stigmatisation.*

However, one anonymous respondent resisted this idea strongly, claiming that this did not constitute an argument in favour of maintaining a certain number of people with Down's syndrome in society.

*It is a nonsense to say that people should bring forth disabled children so that there are enough of them to support a critical mass of effort into absorbing them and caring for them. It is correct to note that if there were only a few dozen Down's children, facilities and amenities for them would be greatly reduced. It is wrong to suggest that, at a societal level this is a bad thing.*

#### **4. Discrimination and rights of disabled people**

As an extension of the previous point, some felt that any increase in terminations of pregnancies affected by a chromosomal disorder constituted, or would give rise to, discrimination. Colette Lloyd said:

*However, an increase in the number of terminations of pregnancies of a child with Down syndrome is a concern of mine as I have a daughter (aged 18) with Down syndrome – and see this as a form of disability discrimination that should no longer be tolerated in today's society. It is unacceptable to the government currently to terminate on the basis of gender, due to gender discrimination issues, and knowing several people who have Down syndrome, I see strong parallels.*

CARE expressed concerns about how this outcome might tie in with objectives for rationing state resources, suggesting that this may involve, or give rise to, discrimination. They cited a UNESCO report which argued that:

*Prevention as a social objective, focused, for example, on reducing care costs for people with congenital conditions or disabilities, cannot be the goal of such screening. That would imply a discriminatory practice that sends the message that these people are unwelcome in society.*

CARE also mentioned the UN Convention on the Rights of Persons with Disabilities and suggested that this may give rise to legal issues with the existence, or development of, screening programmes for genetic conditions. They said:

*Concerns are also raised with regard to the UN Convention on the Rights of Persons with Disabilities (CRPD). In Concluding Comments on reports from Spain and Hungary, the Committee on the Convention called for the action to prevent discrimination within abortion law on the grounds of disability.*

The CMF also thought that disabled people's rights were at stake.

*To introduce a screening test that would result in the increased selective elimination of children with Down Syndrome due to a lack of proper inclusion, accommodation, and support, would violate the rights of disabled persons (under the Convention on the Rights of Persons with Disabilities) and our ethical obligations to disabled people and communities.*

The CMF expressed a more fundamental view that unborn children were persons and as such were, or should be, protected from discrimination by the European Convention on Human Rights.

*We would argue that unborn persons are persons still, and that they should be protected from discrimination by Article 14 of the European Convention on Human Rights, ([Article 14: Protection from discrimination](#) (2016) Equality and Human Rights Commission), recognised by the UK Human Rights Act, 1998.*

The Liminal Spaces Project acknowledged the relevance of human rights to many of the issues raised by NIPT, but argued that human rights law did not apply to fetuses.

*Underlying many of the issues at stake is the question of which human rights are in play, and for whom. It is well established in law that human rights only come into effect once someone is born alive. There is a particular challenge, therefore, if arguing from the perspective of the fetus since it does not have actionable rights as such. This is well recognised in the UK and does not give rise to conflict if, say, NIPT is sought in order to take a more informed decision about pregnancy termination.*

With respect to Down's syndrome specifically, a further issue raised was the severity of Down's syndrome as a health condition and the implications of this for legitimate application of Ground E of the Abortion Act which states that pregnancies may be terminated later than 24 weeks if "there is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped." The CMF argued that there was no such risk in the case of fetuses affected by Down's syndrome with the implication that legal protections for such fetuses were not being enforced.

*To assume that Ground E provision should automatically apply to Down Syndrome is to stretch the application of the Abortion Act (1967) to the point of completely misshaping it. Down Syndrome should not be classed as a 'serious handicap'; and screening for it should only be offered to mothers in*

*order better to prepare them and their families to be joined by a child with special needs.*

Individual respondent, Kay Sammon, also raised the exception to the Abortion Act's 24-week limit.

*There is a further anomaly between improved disabled rights in society, yet a disabled fetus does not share the same right to life as a non-disabled fetus (from 24 weeks) and can be terminated up to 40 weeks (and post-natally in countries such as Holland).*

DSRF UK thought that people with Down's syndrome had a right to comparable investment in medical research, which they said would be compromised by an increase in terminations of fetuses affected by Down's syndrome.

*As a charity that recognises the rights of people with DS to equitable quality medical research we feel that an increase in the number of terminations of unborn persons with DS is a failing of society and the healthcare systems.*

An anonymous respondent, however, felt very differently about the implications of a reduction in the number of births of babies with Down's syndrome for the rights of disabled peoples.

*The reduction in number of Down's births does not give rise to an enforceable right: against whom? All women of childbearing age? There is no such thing as a right to make other people share your disability. Since there is no right to force other people to have children, there cannot possibly be a right to force people to have disabled children.*

## **5. Impact of terminations on women**

The psychological impact of terminating a pregnancy on a woman was raised by some respondents. The CMF, for instance, argued that:

*(Psychological) morbidity following termination of a pregnancy for fetal disability has been shown to be both prevalent and persistent and associated with long-lasting consequences for a substantial number of women.*

The emotional impact of a termination could also extend to other members of a woman's family, the CMF argued.

*Their families are also not immune with even very young children and those sheltered from knowledge of the event showing reactions to their parents' distress and maternal absence.*

The Anscombe Bioethics Centre made a similar point, arguing that women are no better off in virtue of access to abortion.

*Abortion is in no way therapeutic for either mother or child: there is simply no evidence that women are benefited in any way by the particularly painful experience of abortion for foetal anomaly, including for life-limiting conditions.*

## 6. Objections to abortion

Some raised issues relating to abortion itself, and suggested that any increase would, in and of itself, constitute a concern. In addition to the CMF's stated position that unborn persons are persons (see 5, above) the Church of England, Mission and Public Affairs Council said:

*The Church of England combines strong opposition to abortion with recognition that there can be strictly limited conditions under which it may be morally preferable to any available alternative; such alternatives, however, require careful exploration*

A further factor raised by some respondents was the difference between terminating the pregnancy of a fetus affected by Patau's or Edwards' Syndromes and terminating a fetus diagnosed with Down's syndrome. Whilst Patau's syndrome and Edwards' syndrome are of such severity that the Church of England, Mission and Public Affairs Council recognised that in light of the suffering associated with these conditions, "many prospective mothers might feel unable to continue with their pregnancies" the same did not apply to Down's syndrome, they said.

*Down Syndrome however is quite different, with very many individuals living happy and fulfilled lives, giving great joy to their families and contributing richly to the diversity of society.*

The DSA made a similar point, and emphasised the importance of distinguishing issues raised by each of the three conditions from one another.

*The DSA has significant concerns that testing for Down's syndrome is being presented as a 'package' alongside Patau's and Edwards' syndrome. The outcome for babies born with Patau's and Edwards' syndrome is very different than that experienced by children born with Down's syndrome. This association is misleading and could lead to a misunderstanding of the current life chances of people with Down's syndrome and therefore influence the choices women make.*

## 7. Uncertainty about impact on number of terminations

Some respondents challenged the idea that the number of terminations of pregnancies affected by genetic conditions would increase following the introduction of NIPT on the NHS. Clinical Genetics and Cytogenetics, Guy's Hospital said they thought that making NIPT available in the NHS would probably not have this effect.

*Unlikely to result in an increased number of terminations for Patau's or Edwards' Syndrome as these present with ultrasound anomalies and would currently be detected.*

In the case of Down's syndrome, they cited the findings of the RAPID study which suggested that many women would use NIPT to access information that would help them prepare for the birth of a child with extra needs.

*Since this is meant to be offered to women who are at risk of these aneuploidies, they would have had the option of invasive testing anyway, and an increase in the number of TOPs is therefore unlikely.*

Genetic Alliance UK said that there was uncertainty over this outcome.

*It is unclear whether implementation of the UK NSC recommendations on NIPT into NHS antenatal care would lead to a significant increase in terminations of pregnancies with a diagnosis of Down syndrome, Patau's syndrome or Edwards' syndrome. Given that the increased accuracy of the new pathway including NIPT suggests that some 102 additional cases of Down syndrome would be diagnosed prenatally each year, a superficial analysis might suggest that the addition of NIPT to the pathway would lead to a further reduction of births with Down syndrome....However, the RAPID study also found that approximately one third of women with a confirmed positive NIPT result chose to continue their pregnancy, suggesting that the high uptake of NIPT includes women who would like additional information for preparedness and not necessarily for decision making about termination of pregnancy, and who would be reluctant to use invasive techniques because of the miscarriage risk.*

### **8. Aspirations for perfect babies**

The CMF were concerned that NIPT might contribute to the development of societal norms that favour giving birth to 'perfect' children.

*Ultimately, it fosters in society the notion that only the (genetically) perfect, or those who reach a certain arbitrary minimal standard, are acceptable and that it is socially desirable to prevent people with some or all disabilities from being born.*

**Question 4 - Do you think the UK National Screening Committee's criteria for appraising the viability, effectiveness and appropriateness of a screening programme are appropriate for appraising prenatal screening programmes?**

**UKNSC criteria for appraising the viability, effectiveness and appropriateness of a screening programme**

**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 association between the risk or disease marker and serious or treatable disease.
2. All the cost-effective primary prevention interventions should have been implemented as far as practicable.
3. If the carriers of a mutation are identified as a result of screening the natural history of people with this status should be understood, including the psychological implications.

**The test**

4. There should be a simple, safe, precise and validated screening test.
5. The distribution of test values in the target population should be known and a suitable cut-off level defined and agreed.
6. The test, from sample collection to delivery of results, should be acceptable to the target population.
7. There should be an agreed policy on the further diagnostic investigation of individuals with a positive test result and on the choices available to those individuals.
8. If the test is for a particular mutation or set of genetic variants the method for their selection and the means through which these will be kept under review in the programme should be clearly set out.

**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

## **The screening programme**

11. There should be evidence from high quality randomised controlled trials that the screening programme is effective in reducing mortality or morbidity. Where screening is aimed solely at providing information to allow the person being screened to make an “informed choice” (such as Down’s syndrome or cystic fibrosis carrier screening), there must be evidence from high quality trials that the test accurately measures risk. The information that is provided about the test and its outcome must be of value and readily understood by the individual being screened.
12. There should be evidence that the complete screening programme (test, diagnostic procedures, treatment/ intervention) is clinically, socially and ethically acceptable to health professionals and the public.
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.
14. The opportunity cost of the screening programme (including testing, diagnosis and treatment, administration, training and quality assurance) should be economically balanced in relation to expenditure on medical care as a whole (value for money). Assessment against this criteria should have regard to evidence from cost benefit and/or cost effectiveness analyses and have regard to the effective use of available resource.

## **Implementation criteria**

15. Clinical management of the condition and patient outcomes should be optimised in all health care providers prior to participation in a screening programme.
16. All other options for managing the condition should have been considered (such as improving treatment or providing other services), to ensure that no more cost effective intervention could be introduced or current interventions increased within the resources available.
17. There should be a plan for managing and monitoring the screening programme and an agreed set of quality assurance standards.
18. Adequate staffing and facilities for testing, diagnosis, treatment and programme management should be available prior to the commencement of the screening programme.
19. Evidence-based information, explaining the purpose and potential consequences of screening, investigation and preventative intervention or treatment, should be made available to potential participants to assist them in making an informed choice.
20. Public pressure for widening the eligibility criteria for reducing the screening interval. and for

## 1. UKNSC criteria is appropriate for prenatal screening

Some respondents said that the criteria is adequate, responding simply “yes” in a number of cases. ARC’s Jane Fisher replied:

*As a PPI member of the UK NSC, I can attest to the robustness of its processes and the rigour with which the recommendations are considered.*

## 2. UKNSC criteria are not appropriate for prenatal screening

Others however felt that there were issues with the UKNSC criteria and levelled criticisms at its use in the context of prenatal screening. The PHG Foundation said that:

*The PHG Foundation has some general concerns about the current criteria used in the UK by the NSC. In the PHG Foundation document, Genetic Screening Programmes: An International Review of Assessment Criteria we undertook a literature review of genetic screening appraisal as part of the wider NSC review of screening policy. We identified that genetic screening does not fit well with the overall criteria used to appraise a wide range of screening programmes e.g. those for common chronic diseases, which are concerned with reducing morbidity and mortality in the population.*

It was said by individual respondent, Lorna Watson that the criteria take no account of the Public Sector Equality Duty, and should.

*The criteria do not consider the Public Sector Equality Act Duty which is a legal responsibility for the NHS and also applies professionally to public functions of Medical Royal Colleges and Faculties.*

Some respondents cited particular areas of the criteria. Felicity Boardman raised issues with criterion 9, that **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.** This was hard to apply, she said, and raised challenges concerning how to assess whether a better outcome would be achieved, given that terminating the pregnancy was often the principal intervention for a fetus with a genetic condition.

*How do we define ‘better outcomes’ (point 3.9), how do we ever assess if not having that child is a ‘better outcome’ for that screened woman, or indeed that foetus? It is impossible to assess this. Screening decisions that result in the end of life for a foetus are unique and deserve special consideration.*

Colette Lloyd suggested that the UK NSC was not necessarily an appropriate organisation to make judgements about the appropriateness of prenatal screening programmes.

*In short, no. I am not even sure that the NSC with their emphasis on treatable conditions is the correct body to be examining prenatal screening programmes.*

Felicity Boardman made a more general criticism that the criteria are individualistic and should take account of the wider set of interests at stake in screening.

*...Pregnant women, their partners, the foetus, their extended families, communities and wider society are all implicated by screening programmes. The criteria need to take into account the contrasting, and sometimes conflicting, interests of all of these involved parties when evaluating screening.*

Kay Sammon said that the criteria as a whole focus overly on a 'medicalised' view of disability:

*The major concern is that it gives a generalised 'medicalised view' of disability with a focus on individual impairment. We need to urgently challenge these medicalised views of disability ingrained with underlying assumptions about the quality of life and value of unborn disabled babies.*

### **3. Absence of needed criteria**

Some of the criticisms of the UKNSC criteria concerned particular areas where there were perceived to be omissions. For instance, Felicity Boardman argued that whilst the criteria appeal to perspectives on the ethical acceptability of the screening programme to the wider public, the criteria are not sensitive to the views of people with the conditions screened for, specifically.

*For me, this is a striking, and unacceptable, omission. The criteria only make reference to the acceptability of the test to the general public and health care professionals, and yet...people living with the screened-for conditions are directly implicated in the debates around screening. Their lives would be touched in various ways by the outcomes of any such programme and they are therefore an important stakeholder group. Their views need to be considered and valued in the same way as those of the general public and health care professionals.*

Others argued that financial implications for the NHS of implementing a prenatal screening programme should be taken into account within the UKNSC criteria. A member of the BMFMS said:

*[The criteria] should consider the cost implications of reducing the number of babies born with T21.*

### **4. UKNSC criteria are not being applied**

A number of respondents pointed to areas where they thought existing screening programmes did not meet the standards set by the UKNSC criteria. The Anscombe Bioethics Centre said that, whilst flawed, the criteria provided useful guidance in certain areas, but argued that the many actual examples of screening practice do not satisfy these criteria.

*They are insufficient and not wholly satisfactory, but not without some value as they do contain some reasonable requirements: requirements which fail to be satisfied in fact by much prenatal screening practice.*

They also stated that criteria 9, that **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**, criteria 12, that **there should be evidence that the complete screening programme (test, diagnostic procedures, treatment/ intervention) is clinically, socially and ethically acceptable to health professionals and the public** and criteria 13, that **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**, were not met by much existing screening.

*There is for example no evidence that (looking at point 9) the screened individual – whether this is taken to be the baby or the mother – can be the subject of “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”.*

Kay Sammon thought that Criterion 12 (as above) was not met in the case of Down's Syndrome screening:

*There is a body of public opinion including those with Down Syndrome and their families that the tests are socially and ethically unacceptable.*

Future of Down's raised other issues with the application of criterion 12 (as above), since, they argued, the UKNSC was not in a position to make an assessment of whether this criterion was always satisfied, given the potential for broader, long-term societal implications of incorporating novel screening technologies into national screening programmes.

*Constructs within the UK NSC are insufficient to address ethical and societal implications of the technological seep in prenatal screening. Through NIPT, prenatal screening now presents parents with the opportunity to be more discerning about the genetic make-up of their offspring. This has the potential to substantially alter the genetic make-up of society. Whilst conditions such as trisomies and congenital developmental defects will continue to occur sporadically, deselection of foetuses with autosomal dominant or recessive genetic conditions will ultimately lead to genetic homogenisation of society.*

The CMF argued that the second part of criterion 9, that **where there is no prospect of benefit for the individual screened then the screening programme shouldn't be further considered** was not being applied, in the case of Down's, Edwards' and Patau's syndrome prenatal screening, since it is the fetus, rather than the mother, who is screened and does not benefit from the test.

*In cases of trisomies, prenatal screening tests are carried out not to identify individuals with special needs, in order that they may be more effectively treated, but with the expectation of eliminating them from the population.*

DSRF UK agreed with this assessment.

*The detection of unborn persons with DS does not confer ANY advantage on these persons should they be born. There is not protocol to intervene in-utero, for example.*

The DSA suggested that criteria 1, that **the condition should be an important health problem, as judged by its frequency or severity**, did not apply to Down's syndrome, since, they argued, it is not a serious health problem.

*Down's syndrome is not a health condition; it is a chromosomal disorder caused by an error in cell division that results in an extra 21st chromosome. Although certain health conditions are more common in people with Down's syndrome, poor health is not inevitable.*

DSRF UK agreed that "...DS is not a disease as stated by the NSC" as did Colette Lloyd who said that Down's syndrome is compatible with good health.

*Down syndrome is not a health problem. Babies with Down syndrome can be born perfectly 'healthy' from a medical perspective.*

However, the PHG Foundation interpreted this criterion differently, given that, they said, the "scope and purpose of prenatal screening programmes are concerned with offering reproductive choice..." Their view was that:

*Use in prenatal screening presupposes that it is important to be able to offer all pregnant women the opportunity to avoid the birth of a baby who is likely to die or be severely affected by disease that is identifiable before birth. Women would have the choice to terminate the pregnancy. Use of the screening criteria for decision making in prenatal testing implies that any screening programme must satisfy the primary criteria that the availability of this choice (rather than the condition itself) is an important health issue. The psychological, physical and social effects on the mother/parents throughout the pregnancy and in raising the child are thus the outcomes of concern.*

Felicity Boardman raised an issue concerning the application of criterion 11, that **there should be evidence from high quality randomised controlled trials that the screening programme is effective in reducing mortality or morbidity**. It was hard to see how this would be satisfied in the case of prenatal screening, she argued, since one of the metrics associated with effectiveness for NIPT, a reduction in the number of miscarriages, may obtain alongside a correlate increase in the number of terminated pregnancies. This might raise questions about how the significance of these metrics differ from one another, and how to account for each element when appraising the effects of the screening programme in terms of morbidity and mortality.

*How are morbidity and mortality defined in this context? One of the major benefits of NIPT is that it reduces the miscarriage rate (is this defined as morbidity or mortality?), however it may increase the rate of pregnancy terminations in relation to conditions such as Down's Syndrome, Patau's Syndrome or Edwards' Syndrome. This raises the question of how miscarried foetuses are valued vis-à-vis terminated foetuses affected by one of these conditions. If there is not much difference between these two numbers*

*(miscarriage goes down but termination goes up) important questions need to be addressed regarding the moral difference between these two figures. What makes one a marker of success and the other not?*

A more general observation was made by Kay Sammon that there was a lack of consistency in how the criteria was applied to different screening programmes.

*There are inconsistencies in which conditions are screened for, e.g. Cystic fibrosis or Duchenne muscular dystrophy are not, and it should be questioned why a condition with known good quality of life and life expectancy over 60 is targeted.*

**Question 5 - How would you rate the information and counselling currently provided by the NHS to pregnant women and their partners to help them make decisions about currently available prenatal screening (e.g. using ultrasound) for genetic conditions during pregnancy, if you have experience or evidence relating to this?**

**1. High quality information and counselling**

Some respondents said that the quality of information and counselling currently available alongside NHS prenatal screening was high. A member of the BMFMS said that it was “good” and Clinical Genetics and Cytogenetics, Guy’s Hospital said:

*The information given in our trust’s fetal medicine unit by midwives and obstetrician is accurate, although since NIPT is currently only offered privately it is not always discussed as an alternative to invasive testing when women decline this.*

ARC explained that they had been involved in producing the written information provided by the NHS to women considering undergoing NIPT and underlined objectives to ensure it was impartial. They added, though, that however good information provided was, it could be difficult for women and couples to fully process the implications of undergoing screening so early in pregnancy.

*Ensuring that women make considered choices about which screening tests to have is challenging. We were involved in developing the current written information provided by the NHS. Every effort was made to make it balanced and accessible but the reality is that women receive this information early in their pregnancy. At such a ‘tentative stage they don’t always feel able psychologically to fully consider the consequences of their screening decisions. We have to accept that while it is important to make every effort to provide pre-test information (and there is scope for considering new ways of providing it) and to allow time for options to be discussed, there will be significant numbers of women who will opt in hoping to be reassured without giving potential outcomes a lot of thought. It is therefore essential that women who receive a screening result that leads to decisions about further testing are provided with well-co-ordinated care.*

**2. Low quality information and counselling**

Some respondents said that information and counselling made available through the NHS was poor or inadequate. Saving Down Syndrome described information about prenatal screening tests and about disability as “woefully inadequate”. Kay Sammon said that her own research had shown that there were issues with the presentation of information about the realities of raising a child with a disability, specifically.

*The findings of my research ... indicated the majority of parents were largely ignorant about Down’s syndrome (prenatally) and based the decision to*

*continue with their pregnancy on a wide range of personal complex factors... Significantly, there was a gaping hole in the provision of practical, real life information to support and prepare parents for the birth of their child.*

Another area of NHS information and counselling criticised was the information that women and couples are given about the purpose of prenatal screening and conditions it screens for. The Association of Genetic Nurses and Counsellors said:

*There is anecdotal evidence from our membership that patients are poorly informed of what may be detected from current prenatal screening and that the combined screening test for trisomies is sometimes poorly explained to patients. This information has come from observation of antenatal appointments, members attending their own antenatal appointments and receiving feedback from patients.*

It was also said by the Association of Genetic Nurses and Counsellors that not enough was done to convey that prenatal screening was optional for women and couples and not part of standard, maternity care. This could potentially have impacts for patients later, they said, when results are discussed.

*There is also anecdotal evidence that this screening is often presented as routine care, so patients sometimes agree without considering the potential consequences and what they would do if an abnormal result is found. Therefore, a bad news result is a shock and important decisions have to be made in a short timescale.*

DSRF UK criticised the information leaflet given to pregnant women and couples about prenatal screening was inadequate.

*The leaflet about 'Screening tests for you and your baby' is not sufficient to facilitate informed consent. We are often contacted by parents and professionals seeking more information and copies of our new parent booklet despite not having given birth yet.*

### **3. Antenatal Results and Choices (ARC)**

There was some criticism of the charity Antenatal Results and Choices (ARC), an organisation women and couples are advised by the NHS to seek support from when making decisions about pregnancy. Some felt that ARC falsely claimed to be non-directive in their advice. Colette Lloyd, for example, said that ARC information focuses disproportionately on abortion.

*...The NHS currently refer patients to ARC, see previous comment on their activities, their Handbook for all parents on diagnosis of a fetal anomaly, which only discusses termination, and their Continuing Pregnancy booklet, which only discusses the negative consequences of this choice.*

DSRF also said that the information ARC provided was not balanced and suggested that the organisation had a conflict of interests, in virtue of funding it had received from companies that produce NIPT tests.

*The NHS directed use of Antenatal Resources and Choices (ARC) as an information hub for parents is unsatisfactory. It is **not** non-directive in our opinion and should declare that it has received funds from NIPT test producers to every parent who receives advice. This causes harm to professionals and parents because of this competing interest and biased publications.*

Future of Down's suggested that since the ARC began as a body offering support to women who had terminated a pregnancy, it may be difficult for the organisation to completely avoid bias when advising women.

*As a charity Antenatal Results and Choices provide an admirable resource for parents faced with a congenital abnormality. However, despite their evolution throughout the years the derivation of this charity as a support group for parents who have undergone a termination .... cannot be diminished and whilst progress has been made in supporting parents not only after a pregnancy termination but also through the decision making process it cannot be ignored that a potential for bias exists.*

Future of Down's also pointed out that ARC makes use of 'peer supporters' for women who have terminated a pregnancy and for women who have miscarried but none who parent a child with a disability (noting though that ARC instead refer women to charities like the Down's Syndrome Association and the Down's Heart Group). Overall they concluded that:

*...It is questionable whether ARC in its current form is the most appropriate resource for the NHS to direct parents to.*

#### **4. Variability**

A number of respondents said that the quality of information and counselling available in the NHS was variable. A member of the BMFMS said that it was "*generally good but with significant geographical variation*" and an anonymous respondent said it was "*...often good. Sometimes poor.*" Lorna Watson said: "*...anecdotally this is highly variable.*"

DSRF UK pointed towards quality information that women and couples receive about Down's syndrome in the US.

*There are no UK wide materials agreed by both professional bodies and service users. In the USA Lettercare has produced an excellent resource that links up with online videos from the Genetic Support Foundation. These resources could be made available here.*

#### **5. Language**

A related point concerned issues with the language used by healthcare professionals and how this might influence women and couples' attitudes towards screening and termination. An anonymous individual respondent said that:

*Language used both by NSC and staff with respect to 'risk' is highly negative. There is evidence that negative, outdated information is given to parents*

*prenatally that would be entirely inappropriate postnatally. This inconsistency is serious and undermines the Montgomery principles of consent.*

WeLDNurses also said that “...the use of language for framing and describing this area of work was picked up on as significant” agreeing that words such as ‘risk’, ‘sorry’, ‘bad news’ and even ‘screening’ could carry an “...implicit indication of negativity.” They added that:

*This type of discourse could be observed to be implying behavioural ‘nudge’ techniques within public health as observed by Voyer (2015) to encourage behaviour change and influence decision making based on liberal paternalism.*

## **6. Pressure or presumption about screening and termination**

Some raised issues concerning pressure or disapproval to which women and couples might be exposed when being offered information about screening. There were concerns that women may feel that it is expected that they undergo prenatal testing. The Anscombe Bioethics Centre said:

*Staff have anecdotal evidence of women being pressured to receive tests, including a nuchal screening test a particular woman had specifically refused, and where the health professional continued to ‘trail’ information as if hoping the woman would change her mind.*

Future of Down’s said that bias could also be perceived in information concerning decisions around continuing with or terminating a pregnancy.

*...The information and support available exposes a bias towards termination [which] reinforces the perception that prenatal testing is there to provide the opportunity to terminate an affected foetus rather than continue with this addition information about the foetus.*

Saving Down Syndrome shared these concerns.

*It is widely recognised within the Down’s syndrome community that there is a bias towards selective pregnancy termination rather than ongoing support for the pregnancy.*

**Question 6 - How would you rate information and/or counselling provided by the NHS about NIPT available as part of research studies or through the private sector, if you have experience or evidence relating to this?**

***Positive about NHS information and/counselling on NIPT***

**1. Information and counselling is good**

Some respondents said that the quality of information and counselling provided by the NHS about NIPT available as part of research studies or through the private sector was good. An anonymous individual respondent said it was “usually good” and a member of the BMFMS said that “...in the context of research setting I would say reasonably good.”

The Association of Genetic Nurses and Counsellors said that information in certain parts of the NHS was of high quality.

*Within NHS genetics services, we feel patients are generally given comprehensive information including the limitations, possible outcomes and potential consequences of such testing. As an organisation, we do not have evidence or substantial experience of the quality of counselling provided by non-genetics health professionals and private sector clinicians around NIPT.*

The PHG Foundation pointed out that it was hard to make generalisations in this area as the RAPID study provided the principal context in which NIPT had been offered to NHS patients and observed that information and counselling as part of this study had been good. They said that adequately resourcing information and counselling services within the NHS was imperative.

*It is difficult to assess the access to general information on NIPT for patients outside formal research studies or outside areas where an NHS service has been established, but several reviews in the literature highlight the importance of careful non-directive pre- and post-test counselling to avoid undermining informed consent and mitigate against increasing routinisation. The RAPID implementation study group developed materials and provided training sessions for healthcare professionals offering NIPT. The study group also produced materials for parents, which were validated by parent groups and healthcare professionals. Evaluation of parent experiences showed that 88% were found to have made an informed decision, and feedback on the resources was very positive. The study highlighted the critical importance of providing adequate resources in an implementation setting in the NHS to train healthcare professionals to deliver pre- and post-test counselling, and to ensure sufficient time to discuss NIPT and its implications with parents.*

Clinical Genetics and Cytogenetics, Guy's Hospital attested to the quality of the information in their trust, but added that this pertained only to certain conditions.

*The information provided in our trust by the fetal medicine team and/or the clinical genetics team is accurate but currently all that is offered is re aneuploidies and sex chromosomes anomalies.*

### **Negative about NHS information and/counselling on NIPT**

## **2. Quality of information and counselling**

Some respondents said information could be unclear or confusing. Colette Lloyd reported observations from members of her organisation, Down's Syndrome Oxford.

*Further comments from Down's Syndrome Oxford belied a general feeling that information was poor or inadequate or confusing and that there was little support around actually making a decision about testing. One woman was told to have NIPT (that she should pay for) to help the doctor make a decision over treatment...*

It was also said that there were issues with how statistics on the accuracy of NIPT were presented within information and counselling on NIPT in the NHS. Colette Lloyd made this point and also pointed out that the positive predictive value of NIPT for Down's syndrome was also sensitive to maternal age.

*The NHS and private sector providers continue to promote the specificity figures of 99% which are only applicable in the high risk population. This is on advertising and leaflets and from frontline staff. The correct positive predictive value needs to be made clear and also the decreasing reliability of the test according to age.*

ARC pointed out that for some NHS healthcare professionals, making women aware of the private sector availability of NIPT presented ethical issues.

*Some midwives are not prepared to discuss the option when it is not available in the NHS as they view it as inequitable that only women who can afford it can access the testing. We encourage health care professionals to signpost to us so we can discuss the pros and cons and advise women to ask questions of private providers to assess the quality of the service they are offering.*

## **3. Reflex protocol and consent to testing**

Some respondents described problems relating to women's understanding of how and for what their blood would be tested, in the context of NIPT provision. Future of Down's were concerned about the Reflex protocol, in which the blood of women who

receive high chance results from the combined test is automatically subjected to cfDNA testing. This raised issues for informed consent, they said.

*Future of Down's is extremely concerned about the Reflex protocol. The lead investigator on the RAPID protocol demonstrated in a separate study that practitioners may view the consent process for NIPT differently to that for invasive testing and that there is a potential to undermine women making informed choices (van den Heuvel 2010). This study concluded that considering the importance of informed choice in reproductive decision making implementation of any programme based on NIPT should be designed to facilitate this. The REFLEX protocol flouts this consideration by automatically sending blood samples from parents with combined screening result <1 in 800 for NIPT testing. Further concern is raised when even under strict protocolled conditions advice to the parent in question is erroneous. Furthermore, a look at the informed consent for this protocol shows greatly divergent information from UK NSC approved screening materials. (Reflex Informed Consent Form).*

ARC also raised questions about the legitimacy of the Reflex protocol and said that its use had:

*...Involved limited staff training for what is a complex and questionable pathway.*

Saving Down Syndrome raised more general concerns regarding the need to ensure that healthcare professionals discuss NIPT with women and couples and accurately record these discussions. They described the experience of one of their members who had been involved in NIPT screening without their knowledge.

*One of our parents was signed up to an NIPT screen this year, at an NHS Hospital, without even realising it!*

#### **4. Use of negative language**

Another criticism made concerned the language used by NHS healthcare professionals when discussing NIPT with women and couples. Future of Down's described a report from a parent of a child with Down's syndrome.

*I went along to my booking appointment with my eldest daughter who has Down's syndrome and went through all the standard questions. I was then asked whether I wanted combined screening to assess the baby's "risk" of having Down's syndrome or another genetic abnormality. I agreed to the combined screening and was asked whether, should my result be "high risk" if I wanted the new Reflex DNA test that the hospital offered which would test the DNA of the baby from my blood... I received the results of my combined*

*screening test with a letter stating, “we are pleased to inform you that...” which I found upsetting.*

## **5. Variability**

It was also said that information and counselling varied in quality. ARC said

*In our experience this is variable. I was involved in training professionals involved in the GOSH RAPID study and was impressed by how the counselling of potential study participants was conducted. However, this has not been replicated by other studies.*

The quality of information about private sector NIPT was similarly of varying standards, according to ARC.

*When it comes to providing information to women about NIPT provision in the private sector, our understanding is that there is also wide variation.*

A member of the BMFMS said that information and counselling was not consistent throughout the country.

*Improving but could be better. I think we need a nationwide approach. I believe all women with a high risk screening result should be informed of the option of NIPT, even though this is as yet only available privately. There are key facts about NIPT which may not be getting through.*

**Question 7 - How would you rate the information and/or counselling currently provided by private healthcare clinics to pregnant women and their partners to help them make decisions about NIPT, if you have experience or evidence relating to this?**

**1. Inaccurate information about NIPT**

There were predominately negative comments on the standard of information and counselling provided by private healthcare clinics to women and couples on NIPT in response to this question. Some respondents pointed to areas in which it appeared women and couples may have been given inaccurate or misleading information. The DSA said they were concerned that women and couples sometimes came away from private NIPT providers with inaccurate impressions about the test, with potentially damaging consequences.

*One family continued with a pregnancy, having received a negative result believing the test to be diagnostic. Their baby was born with Down's syndrome and the family thrown into turmoil. They felt that they were not provided with enough information about NIPT from the healthcare professional carrying it out and it was not made clear that NIPT is not a diagnostic tool.*

The BMA raised a similar issue.

*We have raised concerns previously about the accuracy and quality of information provided to those seeking testing through commercial companies, particularly given that they have a financial interest in people taking the test. As stated above women and their partners need accurate information so as to weigh up whether they want to take a test. The acceptability of testing direct to the consumer will, in part, depend on the information it aims to provide. Of particular concern are tests where the results can have significant implications, there is a high likelihood for misinterpretation, or there is a risk of harm, severe distress or anxiety to those seeking testing or others.*

The PHG Foundation cited research raising similar issues about womens' understanding of the limitations of NIPT.

*A systematic review by Skirton et al. (Skirton et al. Non-invasive prenatal testing for aneuploidy: a systematic review of Internet advertising to potential users by commercial companies and private providers (2015) Prenatal Diagnosis; 35 (12):1167-75) has identified a number of companies who do not provide adequate information as recommended in professional guidelines, for example, to advise of the need for confirmatory invasive testing in the case of positive results, and the inability of any test to guarantee the health of the baby.*

## 2. Absent information about NIPT

Some respondents suggested that the information and counselling provided in the private sector lacked details on the performance of NIPT as a test for the particular conditions for which it was marketed. One member of the BMFMS said that they were concerned that information about accuracy was not being conveyed.

*I don't know, but I am suspicious. The providers should be talking about positive predictive values, failure rates, delays, other limitations etc.*

It was observed that in the private sector, NIPT was sometimes described simplistically, as a means of reassuring women and couples about the health of their baby. The Anscombe Bioethics Centre claimed this, observing that the potential challenges surrounding a decision to undergo NIPT were sometimes overlooked. They said that the website of one provider of NIPT: pointing out that.

*...Presents it very much as offering reassurance to parents, rather than as something which can itself create serious anxiety and very serious ethical problems.*

Future of Downs also observed that information sometimes focused overly on 'positive' features of NIPT such as ease and safety, and was often presented in promotional format.

*Information on the internet regarding NIPT availability privately focuses on 3 key components; ease, safety and accuracy. The information is almost entirely in advert form; it takes many clicks to drill down through multiple internet pages before you are able to access any factual information about any of the claims made...Information relating to private availability of NIPT is characterised by aggressive advertising practices [and] promises of affirming the normality of a pregnancy.*

## 3. Lack of counselling and support following high chance results

There were reports from respondents of women and couples being inadequately counselled by private providers following high chance results. The DSA described their experience of being approached for support by women who did not fully understand the implications of their high chance NIPT results, claiming there could be:

*Confusion about how to interpret the NIPT result. Whilst the DSA can provide callers with general information about the test, the screening pathway and Down's syndrome, it is for a professional to interpret results. The DSA has received calls from women confused about what the results mean. It is therefore concerned about women who receive a positive result and go on to terminate/continue with the pregnancy, without access to appropriate information.*

The PHG Foundation agreed that there was lack of consumer or patient understanding of the implications of NIPT results, pointing out the potential for women and couples to be exposed to heightened anxiety.

*Anecdotal evidence suggests that women are seeking confirmatory invasive testing in the NHS, and in some instances express concern and anxiety regarding the meaning of results from private providers.*

#### **4. Unduly negative or absent descriptions of genetic conditions**

The absence of balanced, accurate information about the conditions for which NIPT can test was pointed out by a number of respondents. Future of Downs observed that “no information is provided about the conditions tested for” and the Anscombe Bioethics Centre said that where this information was provided it could be negative, or given alongside links to organisations which, in their view, did not provide balanced advice to women about their options.

*While some attempts are made to provide a balanced description of the conditions tested for, descriptions are still too negative, focusing too much on the child's clinical appearance and limitations and with little or no focus on the joy the child could bring to the family, including in cases where life expectancy is very limited. While the sections on particular conditions do provide a link to support organisations, often only one such support organisation is linked to while in every case there is a link to the highly 'pro-choice', pro-screening Antenatal Results and Choices (ARC).*

A related concern was clarity in provider information regarding which particular conditions NIPT could be used to test for and Saving Down Syndrome said that counselling was not perceived by all private providers to be amongst their responsibilities.

*Considering that many Private sector websites are very vague regarding the conditions being screened for, that counselling is not on the agenda (or the website), and that parent stories confirm this, we would rate information and counselling as being poor.*

#### **5. Variability**

Some respondents pointed out that information and counselling was variable and that there were examples of both good and bad practice. ARC said that some private providers made assumptions that women and couples would have informed themselves about NIPT prior to arranging their tests and that the focus of information they supplied therefore tended to focus on presenting NIPT in a positive light.

*We speak regularly to women on our national helpline who are looking for or have had NIPT in the private sector. Our experience is that the quality of pre-test counselling is very patchy. Many clinics assume that women have done their research before coming to them so feel they have little to do in terms of facilitating an informed choice. Information provided by clinics online is more*

*about advertising and promoting the product than promoting informed choice.*

They added though that some private providers had sought ARC's advice on how best to present information on NIPT to women and couples.

*Although I should mention that we have provided advice and training to three private providers to help them make sure they are giving parents balanced and accurate information and that they take holistic responsibility for the whole testing pathway.*

## **6. Poor practice and errors**

Some of the responses to this question contained descriptions of examples of poor practice. The Association of Genetic Nurses and Counsellors explained, for example, that one of their members had counselled a woman who had been given inaccurate test results from a private provider.

*One member described one case where a couple opted for NIPT through the private sector. The couple were initially given an incorrect result (where they were told the fetus had a normal result), then they were re-contacted a few hours later to be informed that the fetus had a sex-chromosome abnormality. Understandably this caused a great deal of distress and mistrust by the couple, and the NHS genetic counsellor involved had to counsel the couple and 'pick up the pieces'.*

Colette Lloyd reported an instance in which woman had been given inaccurate information about that nature of NIPT as a test, and its accuracy.

*I have no personal experience. However a parent from Down's Syndrome Oxford said :“I had the NIPT at a private clinic in Oxfordshire where I was told that the ‘Harmony’ Test was 99.99% accurate (I was told: “the only reason it’s not 100% accurate is because it’s a new test and there isn’t enough data yet to make it 100% accurate”). I now know this is not true.”*

Future of Downs described the experience of a parent seeking NIPT from a private provider, which did not offer or discuss options for counselling at all.

*A parent called Babybond to ask about the processes. They were advised that the test was quick and simple and they could expect a result by email within 7 days, the company representative also advised on other tests that would be included at no extra cost. When the parent asked what happened if it was a positive test, they were told that it most likely wouldn't be but if they were unfortunate they would need to consider their options. The parent asked what would they have to do? They were told it was an individual's decision what to do with the results. So they asked how they would decide. It was necessary for the parent to volunteer the suggestion that they could talk to their midwife. At no point was it suggested that they may need to consider counselling, nor*

*was it indicated that any counselling available.*

It was pointed out by Future of Down's that the Advertising Standards Authority need to receive complaints about the accuracy of information provided on NIPT by private providers or other poor practice before they are able to take action against them.

*Advertising of NIPT in the UK is regulated by the Advertising Standards Agency. There are no separate requirements. Therefore, at present public reporting of concerns is required to effect change in advertising.*

**Question 8 - What information about NIPT and the conditions being tested for do you think should be conveyed to pregnant women and their partners? How do you think that information could best be conveyed and by whom?**

***Information on NIPT***

**1. Strengths and limitations**

It was noted in responses that strengths and limitations of NIPT should be made clear to women and couples before they undergo NIPT. Clinical Genetics and Cytogenetics, Guy's Hospital said that "*information about benefits and concerns/limits*" should be explained and the Association of Genetics Nurses and Counsellors said that "*the limitations of the test*" should be discussed.

Safety and accuracy were considered to be important benefits that women and couples should be aware of. The PHG Foundation said that the women and couples should be told that NIPT is "*safe, convenient and highly accurate when used for the aneuploidy screening*" and a member of the BMFMS said that certain technical aspects of NIPT relating to the performance of the test should be discussed, such as information about the reliability of the tests in different circumstances:

*Positive predictive values, failure rates, delays, other limitations etc.*

Clarity about the fact that NIPT is a test which gives a probability that a fetus is affected by a genetic condition was highlighted as very important by some respondents. Colette Lloyd said that it should be "*reinforced that the test is not diagnostic*" and another member of the BMFMS said that it was important women understood that NIPT must be followed by an invasive test to confirm results.

*Too much to list here but really important they understand it is a screening test and not diagnostic, the detection rates, need for confirmatory invasive test, time scale.*

Lorna Watson said that it should be made clear that NIPT results cannot determine the severity of learning disability or other health problems associated with Down's syndrome in an affected pregnancy.

*It should be clear that the testing gives no indication of likely function.*

And Genetic Alliance UK said that it was important for women and couples to understand that NIPT could not rule out all health conditions.

*Another factor is the need for health professionals to manage expectations of women and their partners, who may have unrealistic expectations of what can be learned in prenatal genetic testing and the limits of our understanding in*

*this area. They may not, for example, understand that testing does not guarantee a healthy baby.*

## **2. Process**

Some of the information that respondents said should be conveyed to pregnant women and their partners concerned the process of undergoing NIPT itself. This should be discussed explicitly with women and couples and should cover a number of areas including, Felicity Boardman said, “*what the screen involves, what is being tested for, timeframe for results*” as well as “*information about how results will be communicated (and by whom)*”. This was a topic the DSA agreed women and couples should be able to discuss beforehand with a healthcare professional.

*Discussion with a healthcare professional before screening about how they will receive their test results.*

### **Information about genetic conditions**

## **3. Medical and social information**

Some respondents said it was important to make sure women and couples had access to up to date information about the nature of tested-for genetic conditions for those people who have them and their families. ARC said all providers should direct women and couples towards reliable sources of this kind of information and advice on the relevant conditions.

*Private providers should at the very least signpost or have web links to accurate sources of information on the common trisomies.*

Some respondents felt that duties in this area were stronger and that providers had responsibilities for giving women and couples this information themselves. Future of Downs said that medical information about the conditions tested for should be included so that parents “*...understand the medical aspects of a condition and the possible outcomes*” and Felicity Boardman said that a wide range of information about the medical features of the conditions should be given to women and couples:

*The prevalence of the conditions (i.e. likelihood of screening positive for it), the aetiology of the conditions (and if they are inheritable whether and how others in the family might be affected), the range and type of clinical features associated with the conditions, how the conditions are confirmed/ diagnosed after the birth of the child, any possible treatments or options for palliative care and when these would likely occur, the range of prognoses associated with the condition, lifespan implications...*

The DSA said that information provided should include “*...balanced accurate up to date information about Down’s syndrome*” and Lorna Watson argued that as well as information about medical problems associated with Down’s syndrome, accounts of

the many positive aspects the lives of people with Down's syndrome's should be provided.

*Information should include the range of achievements and inclusion possible for people with Down's Syndrome, as well as likely differences and challenges... Positive achievements range from skiing, modelling, professional photography, giving talks at conferences, working as a zumba instructor, being awarded MBE etc.*

#### **4. Sources of support**

Some respondents said that including information about sources of support was important. The DSA said that "...being made aware of the DSA" would be of use to women and couples considering undergoing NIPT and Felicity Boardman said that "sources of support available to families living with the conditions and the contact details of the main support or advocacy group to offer insight into daily life with that condition" should be made known. Lorna Watson agreed and added that these sources should include parents of children with the conditions tested for.

*It should include the support available through early intervention, inclusive education, third sector organisations and other parents.*

Genetic Alliance UK argued that support should be "sensitive and non-directional emotional and psychological".

#### **How should the information be conveyed and by whom?**

#### **5. Specialist healthcare professional expertise**

It was said by some respondents that it was important that whoever relayed this information should have a good understanding of NIPT. Felicity Boardman said that information "...would be best conveyed by a healthcare professional". A member of the BMFMS said that specialist midwives should be involved and noted an issue with regional variations in arrangements for maternity care.

*Seems obvious but needs to be communicated by people who know what they're talking about. In the medium term this is likely to involve midwives with specialised fetal medicine experience. Note in Scotland we don't have an established system of screening coordinators.*

The PHG Foundation agreed that it would be appropriate for midwives to have these discussions with women and couples.

*Midwives are in general best placed to offer this information rather than GPs (who typically do not have the requisite training).*

Felicity Boardman said that where information about genetic conditions was being relayed, a mixture of different expertise would be useful.

*This information would be best conveyed by a range of people, health care professionals (clinical implications, prognosis, diagnosis and treatment), support and advocacy groups to offer daily living information.*

It was stated by one respondent that the role of the person providing information was less important than the fact that they be adequately trained.

*Staff administering NIPT would have to understand all the issues given above and would be required to be highly trained. It does not matter who administers, what matters is that they understand the issues we outline above and can work towards the most positive outcome.*

## **6. Genetic counsellors**

A number of respondents highlighted the need for genetic counsellors, specifically, in discussions with women and couples considering undergoing NIPT. Clinical Genetics and Cytogenetics, Guy's Hospital said that "*this information should be conveyed by clinical geneticists/genetic counsellors*" and DSRF UK concurred, adding that NIPT would present challenges even for highly skilled professionals.

*It is our belief that a specialist role (i.e. genetic counsellor) will be needed to facilitate the workings of antenatal care in the near future when a woman presents with a privately obtained NIPT report. However even the best Genetic counsellor will struggle to answer the questions NIPT reports will raise, questions that even world class researchers argue about.*

Consultation with a genetic counsellor was also ARC's recommended option.

*When it comes to offering NIPT for a wider number of genetic conditions we would like to see providers advising caution and offering a consultation with a qualified genetic counsellor to help parents gain a better understanding of what is being tested for and what results might mean for them.*

Future of Down's also stressed the importance of involving genetic counsellors, pointing out that the skills required to adequately counsel and support women and couples considering prenatal screening were not necessarily ones that doctors and midwives would have.

*Prior to embarking on NIPT prospective parents must to be able to access a dedicated genetic counselling service. This affords respect to the parents and the difficulty afforded by the decision making process. It also respects the technical expertise of appropriately qualified counsellors to guide parents through the decision making process. It would be absurd to expect a genetic counsellor to deliver a pregnancy or conduct fetal surgery. In much the same way it is absurd to expect a midwife, or fetal medical consultant to be sufficiently qualified to be able to counsel parents on the decision making process in respect of continuing or terminating a pregnancy based on the possible presence of a genetic condition.*

## 7. Specialist expertise and personal insight

A further point made was that information on conditions should be relayed to women and couples by organisations with specialist expertise on those conditions. The Anscombe Bioethics Centre said:

*Any information on a particular condition (as opposed to information on NIPT which we do not support as a generally available test) should be provided by a charity dedicated to providing support for individuals with the condition. For example, information on Down's should be provided by one or more of the organizations in this country that support children and adults with Down's and their families and carers.*

Rachel Siden said that opportunities to speak to others who have had NIPT would be helpful for women and couples considering undergoing NIPT.

*Sharing stories from other patients who used NIPT and how getting these tests either empowered or challenged them could help pregnant women and their partners fully envision what these tests could offer to help them decide what is best for them.*

A member of the BMFMS argued that information should be provided by someone with no commercial interest in the sale of NIPT.

*Always better that info like this is provided by someone with no financial motivations.*

### ***How should information be shared?***

## 8. Face-to-face discussions and written information

Some respondents argued that it was important for information to be conveyed in face-to-face conversations with women and couples in addition to giving them written information. These discussions should supplement leaflets or other written materials given to women and couples. The Association of Genetic Nurses and Counsellors said that:

*Information should be conveyed verbally by the antenatal team and good quality written information, from a reputable source, should be given to the couple.*

The DSA also said that their members had reported that discussion of the issues relating to NIPT in-person would be beneficial.

*Information delivered in person, ideally by someone they know, trained in delivering the information in a supportive non-directive manner.*

The need for adequate time for professionals to discuss NIPT and the conditions for which it tests with women and couples was mentioned by some respondents. The PHG Foundation said that women should have “*sufficient time for reflection*” and Saving Down Syndrome said:

*Information about which conditions are to be tested for has to be provided in black and white, or colour ideally, for users, and they have to be given time to understand what these conditions are, before consenting to testing.*

## **9. Format of information**

The particular means of conveying information could be important, some respondents said. Felicity Boardman argued that more modern formats, such as website and video hosting sites, should be utilised and would enable women and couples to more easily access to the insights of parents of children with genetic conditions.

*You Tube videos and a website with some of this information (especially the social support and daily living information) could be used as a supplement to the above information. This would allow prospective parents to virtually ‘meet’ someone with the condition, without the (potential) tensions involved in actually meeting them in person, although options for this could be made available through the support group and would have to be carefully thought out to support the emotional well-being of both the disabled person and the prospective parents.*

She also observed that what is appropriate might depend on what point in the screening process a woman or couple is in.

*The level and type of information prospective parents will require is likely to be different according to their stage in the screening processes*

DSRF UK said that different media for conveying this information were important, adding that it should be available in different languages.

*Information delivered: methods as detailed above in multimedia format in multiple languages.*

## Question 9 - What might be the implications for the NHS of increasing numbers of pregnant women purchasing NIPT through the private sector?

### 1. Potential for increased numbers of invasive diagnostic procedures

Some were concerned that the number of invasive tests the NHS provided may ultimately go up as a result of women receiving high chance NIPT results in the private sector. ARC pointed out that women may choose to undergo NIPT privately for a range of conditions, many of which are less reliably tested for by NIPT, and might want an invasive test afterwards.

*A particularly unwelcome possible implication is that more invasive tests are actually carried out than currently if women choose to have testing for a wider range of conditions including microdeletions and microduplications and so the screen positive rate increases.*

Members of the BMFMS also raised concerns about the variety of results that NIPT might yield when accessed through the private sector. One member raised a concern about:

*...The opportunity for private clinics to test for genetic mutations without a clinical genetic support network to discuss results in an informed way. I can just imagine women turning up to a random DGH antenatal clinic with a complicated genetics result and there being no system in place as to how to provide reliable information.*

A member of the BMFMS pointed out that a number of these extra cases might be women who were found to be 'low risk' by NHS combined screening who receive a high chance NIPT result in the private sector and turn to the NHS for diagnostic tests.

*We might see an increase in the number of invasive tests if women who are truly low risk choose to undergo NIPT (where the PPV will be significantly lower, particularly for T13/18 and 'common' deletions). Unless there are well described pathways, the NHS may be faced by women struggling to access IPT through the NHS when private results have been positive.*

The PHG Foundation raised concerns about whether the NHS had adequate resources to meet these potential needs.

*...There might be a lack of capacity within the NHS to meet the demand for invasive testing, and subsequent management for these women.*

Though another member of the BMFMS suggested that in the short term there might be "lowering invasive testing rates."

## 2. Follow-up care and counselling after privately provided NIPT

A number of respondents pointed out that women who had sought NIPT in the private sector may need recourse to NHS healthcare professionals after receiving their test results. Lorna Watson said that this had potential to “*increase NHS workload.*”

ARC agreed that women may turn to the NHS for further support after having undergone NIPT in private settings and called on private providers to provide more and better follow-up services, such as invasive testing.

*As with other areas of private health care the danger is the NHS ends up ‘picking up the pieces’ when women receive worrying results.... We believe it is incumbent upon private providers to take responsibility for the whole pathway and be in a position to offer quality assured diagnostic services when necessary. This is the approach taken by the Fetal Medicine Centre in London (Prof Kypros Nicolaides’ private clinic).*

Clinical Genetics and Cytogenetics, Guy’s Hospital said that “...*women with high risk / inconclusive / failed NIPT results may require additional counselling / testing by the NHS*” and the Association of Genetic Nurses and Counsellors agreed.

*An increase in workload as pregnant women may then present to NHS services with high risk results and NHS professionals will then need to manage these patients appropriately.*

Felicity Boardman said that counselling on the nature of conditions NIPT tests for might be a particular area where the NHS might become involved.

*The need to re-educate women about the conditions and NIPT. There is no regulation of the information provided to women in the private sector, and can lead to situations where pregnant women are being told ‘the worst case scenario’ in relation to their reproductive risks in order to sell more screens. The NHS might be called upon to balance this information.*

DSRF UK said that this might present particular challenges for the NHS in cases where NIPT results reveal a wider range of genetic information about a fetus, since NHS healthcare professionals would not necessarily have the expertise to fully interpret or explain these.

*This will lead to a greater unwieldy burden upon the NHS. There is a lack of skills, time and scientific knowledge to interpret the reports as they include more and more detail. Patients will demand invasive testing to confirm whatever findings they fear on the report e.g. APO E gene for Alzheimer’s.*

### **3. Potential for increased numbers of abortions**

The Anscombe Bioethics Centre pointed out that women who had undergone NIPT in the private sector and were ultimately diagnosed with a fetal anomaly, and who wanted a termination, would be likely to seek this in the NHS. Colette Lloyd said that “*women may arrive at the door of the NHS following a high risk NIPT demanding an amnio or a termination*”. She said that:

*One very likely implication would be an increase in NHS-funded abortions. The NHS may also face pressure to compete with the private sector in the provision of tests which provide no benefit to women or their unborn children.*

The DSA said that any increase in the number of abortions should be monitored.

*It is possible there might be an increase: In the request for terminations which will need to be monitored.*

### **4. Pressure to make NIPT more widely available**

Some respondents suggested that the proposed arrangements for NHS prenatal screening might be challenged by patients as awareness of NIPT and its use to test for more conditions grows. It was said by one member of the BMFMS that there was potential for this trend to “*undermine the screening programme...*” and Felicity Boardman said the NHS may experience pressure to widen the range of conditions for which it used NIPT to test.

*Pressure to make NIPT available on the NHS for an increasingly wide range of conditions. The private sector typically sells screens for a wider range of conditions than currently available on the NHS. This might lead to calls to expand NHS screening in line with what is being sold in the private sector.*

### **5. Equity**

The PHG Foundation raised concerns about fairness and argued that more women “*...purchasing NIPT through the private sector could result in growing inequity of access.*” One anonymous respondent said that these concerns also had reputational issues for the NHS.

*Poor for the reputation of the NHS in providing care free at the point of access and it would be tangible evidence that the NHS service is failing to meet the health needs of the population. Women having to pay for this service would accentuate health inequity with the more vulnerable members of society having reduced choice and potentially increased risk to their pregnancies. There would also be a lack of continuity of care with fractured pathways and a risk of poor communication.*

On the other hand, a member of the BMFMS suggested that the availability of NIPT in the private sector might lessen pressures on NHS resources by reducing the number of women who would want to undergo NIPT in the NHS (adding that it might create problems for the NHS in other ways).

*This could help to limit the resource drain on the NHS, but may undermine the screening programme.*

**Question 10 - What benefits and concerns might be raised if pregnant women were able to purchase NIPT directly from providers (e.g. where a kit is sent to the pregnant woman in the post), rather than through a healthcare clinic following a face-to-face consultation?**

**Benefits**

**1. Access and convenience**

Some respondents pointed out that it might be easier for some women to access NIPT through direct-to-consumer providers. Felicity Boardman said:

*There might be shorter waiting times for results within the private sector... Pregnant women can undergo the screen relatively easily and in the comfort of their own home.*

PHG Foundation raised these points too, pointing out there might be a range of reasons why women may be unable to visit a clinic or hospital. Direct-to-consumer tests might make access to NIPT more equitable.

*NIPT testing would be more accessible to a wider range of women who perhaps could not attend a healthcare clinic. This might include women who lived a long way from a clinic, or those who were fearful of stigmatisation (either from their own families or the wider community); this might include very young women (including children below 16). Provision of these technologies on a direct-to-consumer basis would help to ensure that women had equitable access to these technologies, an important principle which should underpin the provision of publicly funded health services.*

It was pointed out that women would be able to access NIPT through direct-to-consumer providers earlier than through the NHS.

*The screen can be done from ten weeks of pregnancy. Within the NHS, NIPT is only offered after the 12 week scan.*

**2. Cost**

Some respondents pointed out that the direct-to-consumer tests would be cheaper than those provided by private hospitals and clinics, which would benefit some women and couples. A member of the BMFMS said that "...benefits are price and availability". The Association of Genetic Nurses and Counsellors suggested that there might be cost savings for the NHS, in virtue of reducing demand for NHS provided screening services.

*...May reduce use of antenatal screening services of NHS.*

## Concerns

### 3. Technical understanding of the implications of results

A common concern related to how well women and couples would be able to understand the information provided by their NIPT results, if these were received outside the context of healthcare professional support. The BMA argued that there were particular concerns about direct-to-consumer testing where “*there is a high likelihood for misinterpretation*” and the CMF warned about the risks of women and couples accessing “*...a mass of complex and confusing data without a context in which to discuss meaning and relevance*”.

BioCentre said this issue should be mitigated by the provision of quality patient information.

*This raises concerns about misinterpretation of test results or patients not fully comprehending the limitations and implications of testing<sup>3</sup>...The need for clear and accessible information to support such tests and guidelines then becomes critically important.*

A member of the BMFMS said that discussions with women and couples about the meaning of NIPT results prior to their choosing to undergo testing would help to address this problem.

*Establishing an awareness of the implications of results before testing is the clear concern and women turning up with results where the implications haven't been considered beforehand.*

Though it was added that this was “*...probably not a major problem*”.

The Anscombe Bioethics Centre agreed that this was a concern, pointing out that one implication of this is that women may consequently make decisions about their pregnancy and take actions based on partial understanding of the information.

*We fear...that if NIPT kits were available directly from providers then there would be an increased risk of women misinterpreting results and/or acting impulsively, leading to increased termination of disabled and of able-bodied children.*

The CMF agreed that there were concerns about “*...The danger of knee-jerk requests for abortion*” and Clinical Genetics and Cytogenetics, Guy's Hospital also raised the “*risk also of increase number of TOPs carried out for so-called “social” reasons if NIPT results not discussed through healthcare services.*”

### 4. Absence of support or counselling

A related concern was for the lack of support for women and couples accessing NIPT through direct-to-consumer providers. A member of the BMFMS said that there might be “*poor levels of support*” and the Association of Genetic Nurses and

---

<sup>3</sup> See BioCentre's full consultation response for references.

Counsellors said that the fact that there would be “no counselling involved” was a concern. Future of Down’s said:

*Future of Down’s considers direct to consumer NIPT to leave prospective parents in a vulnerable position without the support of an appropriately qualified individual to guide them through this difficult process.... for those prospective parents where a condition is diagnosed, or where test results are inconclusive, false positive/negatives or show incidental findings availability of DTC NIPT poses a risk to their emotional and mental wellbeing at a time of heightened anxiety such as all parents experience during pregnancy.*

The CMF also expressed this worry about a lack of quality counselling.

*Absent or cursory pre-test and post-test counsel and information by trained staff.*

DSRF UK said that this was especially concerning in cases where women receive high chance NIPT results.

*The most vulnerable group is those receiving a result with a high chance the baby has DS/other issue as they will be alone and not keyed into antenatal system of care.*

And Felicity Boardman added that women and couples might also be in particular need of advice and guidance if they receive inconclusive NIPT results.

*The lack of a face-to-face consultation might leave women feeling under-supported if a high risk or inconclusive result is returned.*

Counselling and support was particularly important, Rachel Siden said, since tests may have been vigorously promoted by the private companies selling them.

*The face-to-face consultation can allow clinicians to offer their patients a clearer picture of how NIPT could affect them based on past experiences with other patients. Buying a test independently, especially if these tests were marketed in convincing or misleading ways, could leave pregnant women very unprepared for an unexpected test result.*

## **5. Quality control and test performance**

Some respondents were concerned about the quality of tests provided this way. A member of the BMFMS said that there was “no governance or quality control” and a “lack of data on effectiveness”. Genetic Alliance UK said:

*We are already seeing commercially available genetic testing making claims well beyond what is possible at current levels of scientific and medical knowledge, and purchasers being sent massive amounts of information without any context or explanation.*

There might also be issues with the process by which NIPT results are generated, the PHG Foundation said:

*It might be more difficult to ensure that blood samples are of a high enough standard for accurate, consistent testing results.... More likelihood of sample*

*mix-up or contamination than where samples are collected by health care providers.*

The BMA expressed a range of worries in this area relating to the nature of information provided to women and couples.

*The acceptability of testing direct to the consumer will, in part, depend on the information it aims to provide. Of particular concern are tests where the results can have significant implications, there is a high likelihood for misinterpretation, or there is a risk of harm, severe distress or anxiety to those seeking testing or others.*

## **6. Legal issues**

Some concerns related to the legal and regulatory background, and areas where women accessing direct-to-consumer NIPT tests might be unprotected. For example, Felicity Boardman said that there was no regulation of the information that is provided to women about NIPT in these circumstances.

*The information provided to pregnant women is not regulated. The information about the conditions and risks to the foetus might be entirely inaccurate.*

The fact that some direct-to-consumer tests may take place outside of the UK may leave women without legal protections regarding the storage and transfer of their blood samples, it was noted. The DSA said that:

*There would be no protection for women/families as testing would be carried out across borders and regulation therefore impossible to enforce.*

## **7. Costs to the NHS**

As with more general concerns about private NIPT provision, some respondents pointed out that direct-to-consumer tests created the possibility that women and couples, in the absence of support, would approach NHS healthcare professionals for advice and guidance on the meaning of their results. This would have implications for NHS resources since women may “...need to be followed up in NHS clinics where the mess will need to be unpicked”, a member of the BMFMS said. The BMA argued:

*The availability of NIPT directly to consumers may also represent a challenge to NHS staff. Where the tests are for conditions or predispositions which would not normally be tested for within the normal screening or testing pathway or if they produce findings which are of unknown clinical utility or which only offer a risk of susceptibility to disease, patients may look to NHS clinicians or other staff to help with the interpretation of results and provide guidance. This can create a pressure on limited resources which would not otherwise have existed. Given the broad spectrum of information it can yield, this will be a particular problem if the technology for sequencing the fetal genome becomes commercially viable.*

The BMA explained that they had asked the Government to provide advice on direct-to-consumer testing, in which the benefits of discussing options with a healthcare professional beforehand could be set out.

*To help address concerns that patients may be accessing testing based on imperfect knowledge and understanding of its implications the BMA has in the past called for official government websites which would provide accurate information about testing to consumers. (British Medical Association (2012) [Medical Ethics Today: The BMA Handbook on Medical Ethics and Law](#) Wiley: Chichester, p.395) This could include the benefits of prior discussion with a health professional and the standards they should expect to receive from companies. We are aware that the National Screening Committee has recently produced general guidance for individuals who are considering having a private screening test. International rules governing the quality of information could also be explored, similar to the agreement within the pharmaceutical industry about the type of product information that should be provided with medication.*

## **8. Conflict of interests for providers**

Some respondents suggested that since for-profit providers were involved, there was a risk that patient understanding would not be the priority for manufacturers of direct-to-consumer NIPT tests.

*Commercial pressures may prevent accurate and independent information from being provided.*

The DSA agreed that “*Women would be in danger of exploitation for commercial gain*”.

## **9. Sex selection**

The PHG Foundation pointed out that some women and couples may use the results of direct-to-consumer NIPT to inform decisions about terminating a pregnancy based on fetal sex.

*Women might be more likely to be pursuing the test for their own reasons (such as social sex-selection).*

**Question 11 - A small proportion of NIPT tests will return an inconclusive result, even if repeated. How should healthcare professionals, both in the NHS and in private clinics, deal with inconclusive results?**

**1. Discussion of possibility of inconclusive results prior to test**

Some respondents said that it was important that the possibility of receiving an inconclusive result should be discussed with women and couples before they undergo NIPT. The Association of Genetic Nurses and Counsellors said:

*This should be raised as a possibility with the pregnant woman and her partner before testing is carried out in both the NHS and private sector.*

The DSA agreed, arguing that:

*Healthcare professionals should be in a position to ensure women are aware of the possibilities prior to testing and know what their options are following any result.*

Future of Down's made the same point, and observed that this created the possibility of delays in testing for women and couples, when they receive an inconclusive result.

*Future of Down's considers it essential that parents are fully informed of the potential for inconclusive results and the likelihood of this occurrence prior to agreeing to NIPT. It is important that a parent should be aware of the potential need for retesting which could again lead to an inconclusive result and must consider the potential delays incurred to decide whether this is acceptable for them.*

And the CMF agreed, stating that “*delays between first and subsequent tests will clearly be anxious times for patients and their families*”. The CMF reiterated the point that women should know in advance that an inconclusive result is one possible outcome of undergoing NIPT, linking the issue with more general concerns that women and couples may perceive NIPT to be diagnostic.

*If it is offered, healthcare professionals must make clear before screening that the new test is not conclusive – it is an advanced screening test but not a diagnostic test. There is already a general perception that the NIPT test at ten weeks will replace other testing because of its reliability. False-positive rates for NIPT are in the order of 0.1%–0.2%<sup>4</sup>.*

**2. Next steps following inconclusive results**

Respondents described the different options that women should be given following an inconclusive NIPT result. The option to follow with an invasive diagnostic procedure should be discussed in the event that a woman receives an inconclusive

<sup>4</sup> See CMF's full consultation response for references.

result, it was said by some respondents including one member of the BSFMS who said that healthcare professionals should “offer IPT as we do now, based on a risk threshold for combined screening.” The Association of Genetic Nurses and Counsellors said that invasive diagnostic tests should be discussed.

*Further options, such as invasive testing, should be offered after inconclusive results.*

Genetic Alliance UK said that women should be given the options of opting out of screening, repeating NIPT or undergoing invasive testing.

*If women receive adequate information and appropriate decision making support both before and after the test, inconclusive findings can be explained to the woman, and she can be supported to make an informed decision about whether to halt testing, repeat the test, or take an invasive test<sup>5</sup>.*

The PHG Foundation agreed that women should be given the option to undergo NIPT again, arguing also that further research into the causes of inconclusive results should be ongoing.

*Given the potential for anxiety arising from inconclusive results, there should be clear protocols on offering repeat NIPT or invasive testing, and ongoing efforts to improve knowledge regarding the reasons for inconclusive results as understanding of the assay develops.*

And ARC specified that in the private sector, a re-test should be offered to women at no extra cost and further advice offered in the event of a second inconclusive result.

*In the private sector at the very least there must be the provision of a second test free of charge. If there is a second inconclusive result it is important the woman involved has access to specialist advice, most likely from a fetal medicine specialist unit where there is also prenatal clinical genetics expertise.*

Clinical Genetics and Cytogenetics, Guy’s Hospital said that management should depend on the particular kind of inconclusive result that a woman received, pointing out that an invasive diagnostic procedure might be appropriate to test the fetus, but that tests for the mother may also be appropriate.

*There are different types of inconclusive result so this would depend on which chromosome and aneuploidy is involved. Invasive testing to assess the fetus, parental testing for CNVs, maternal testing for mosaicism/malignancy are possible ways forward, as is frequent ultrasound monitoring.*

The Anscombe Bioethics Centre, however, argued that invasive diagnostic procedures should not be the default next step.

---

<sup>5</sup> See Genetic Alliance UK’s full consultation response for references.

*Unless there is an urgent need to diagnose the condition to treat the unborn child, the very real risks of follow-up invasive tests such as amniocentesis should not be contemplated. (As a comparison, few of us would accept a test for newborn babies which resulted in death for one baby out of every hundred newborn babies tested, unless the medical need for the test was very urgent indeed).*

### **3. Inconclusive results indicative of a chromosomal disorder**

One respondent pointed out that current research into the causes of inconclusive results suggested that this was sometimes connected to the presence of a chromosomal disorder in the fetus, which should be taken account of in post-test counselling. ARC said:

*There remains uncertainty as to what an inconclusive result might mean and whether it may sometimes be indicative of an increased chance of trisomy being present. So women need careful counselling in this instance.*

### **4. Regulation and oversight**

Future of Down's said that there was a need for regulation and evaluation which should include reporting of inconclusive results, as well as incidental findings.

*Future of Down's is...concerned that due to inadequacies in regulation of NIPT the true scale of these incidental findings is underreported and thus insufficiently characterised. Appropriate regulation should require thorough evaluation and reporting of incidental findings, inconclusive results and findings of false positives and false negatives.*

DSRF UK thought that the possibility of inconclusive results meant that a "robust review of the ethics must be undertaken to explore these issues."

**Question 12 - What issues are raised by incidental findings that can arise following NIPT (such as genetic abnormalities or cancerous cells in the pregnant woman), both in the NHS and in private clinics?**

**1. Discussions prior to consent and testing**

Some respondents said that the possibility of incidental findings should be discussed with women before they undergo testing. A member of the BMFMS stated that “...*this is about pre-test education and consent, and having clinicians who know to expect these and how to handle them.*” Future of Down’s agreed, arguing that the probability that an incidental finding will be made should be addressed.

*Future of Down’s considers it essential that parents are fully informed of the potential for incidental findings and the likelihood of this occurrence and its implications prior to agreeing to NIPT.*

Genetic Alliance UK pointed out that issues relating to incidental findings were well understood in the context of other kinds of testing and argued that existing research in the area should be utilised to develop good practice policies on incidental findings in NIPT.

*This is far from a new question - all medical investigations come with a chance of incidental findings. We would suggest that the screening programme learn from the extensive work done on this subject for genetic testing in research or for diagnostic purposes<sup>6</sup>. We understand that best practice involves discussing the possibility of incidental findings with the woman while taking consent to testing, and recording her preferences for disclosure, particularly as relates to non-medical findings such as relating to parentage.*

The PHG Foundation agreed that discussions involved in seeking consent should include the frequency of incidental findings, the circumstances in which they would be reported and possible treatment options, if necessary.

*...If NIPT is to be used more widely, the consent process should include a discussion of any additional or incidental findings that might be generated by the test; their frequency; the extent to which they might be reported and any additional actions or management that might be suggested or required if they are detected.*

Clinical Genetics and Cytogenetics, Guy’s Hospital agreed that the possibility of such findings should be discussed before women undergo NIPT and observed that women may otherwise receive information that they do not want.

---

<sup>6</sup> See Genetic Alliance UK’s full consultation response for references.

*Ideally these findings would be part of the informed consent but in reality such cases are rare and in practice this information may not be provided prior to testing. Women may not want to be provided with the information but if this isn't detailed in the consent are likely to be provided with the information.*

## **2. Need for further support when incidental findings are disclosed**

Some respondents argued that in the event that women are provided with information about incidental findings there must be adequate support made available for them. ARC stated that appropriate advice on the implications of the findings should be offered to women in these circumstances, whether NIPT is accessed privately or through the NHS.

*In both sectors women will need to know that this is a rare but possible outcome and specialist advice should always be available to help her understand the possible implications in her individual case.*

The Association of Genetic Nurses and Counsellors also said that support should be accessible to women who receive incidental findings following NIPT and pointed out that this might need to be accessed through the NHS, even in cases where NIPT has been sought from a private provider.

*Unexpected potentially life-changing information may be received by the pregnant woman and her family as a result of such incidental findings. There may be a need for downstream genetic counselling or treatment may therefore arise, potentially needing to be provided by NHS services after testing in the private sector.*

Felicity Boardman pointed out that increased stress and anxiety was particularly problematic for women during pregnancy, meaning psychological support should be available.

*Pregnancy can be a challenging time, particularly for a couple who might have already received a high risk screening result (hence the decision to use NIPT). A serious incidental finding might cause considerable (and additional) emotional distress to the pregnant woman, which, in turn, could affect the well-being of the foetus who are responsive to stress hormones.*

## **3. Information about the woman's health should be disclosed to her**

A number of respondents suggested that incidental findings should be made known to women, whether or not this had been previously discussed. The CMF said:

*Test results should not be withheld from the patient, even if some studies have shown that such early detection does not always improve patient outcomes<sup>7</sup>.*

---

<sup>7</sup> See CMF's full consultation response for references.

And they added that there should be “*onward referral to appropriate specialists at the first opportunity, to minimise the length of anxious waiting times*”.

The Anscombe Bioethics Centre said that information about maternal cancer, specifically, should be disclosed in view of the fact that such findings would give rise to serious health implications (though argued also that some incidental findings should not be disclosed).

*Should tests be carried out (although we do not recommend NIPT as a standard procedure) with the unexpected result that cancer is discovered, the cancer should of course be dealt with appropriately in a way that targets the cancer, not the baby... There is a case for proactively offering this information to the woman, even if the finding is incidental, since the need for uncontroversially beneficial treatment may be urgent.*

#### **4. Issues with disclosing information to the woman**

Some respondents raised concerns about relaying incidental findings to women and the potential risks involved. One member of the BMFMS said that samples should be tested only for the variations for which a woman has given consent to have her fetus tested for (meaning that incidental findings would not arise).

*My initial reaction is to only test what you're looking for i.e. answer the question or questions. If the question is “are there signs of a trisomy” then the answer should be yes or no and any other suspicions not reported until there are robust data as to what these result actually mean.*

The same respondent pointed out that some incidental findings can give rise to anxiety about health but yield no useful information in terms of care management.

*There are several examples such as CA 125 testing in pregnancy or PAP prostate acid phosphatase in asymptomatic men where the result causes panic but no useful intervention.*

Felicity Boardman agreed that there was an issue with relaying incidental findings.

*Which incidental findings should be reported? Some findings will be of clinical relevance, whereas others will not be. Concerns arise around which are relayed back to the pregnant woman and which are not.*

The BMA raised the same concern about causing unnecessary anxiety in women, as well as over-diagnosis and over-treatment. They recommended that further research into incidental findings associated with NIPT be undertaken.

*Finding additional, unsought or unexpected information through genetic testing is a longstanding issue which is likely to become more problematic if sequencing develops, more genetic information is yielded and genome sequencing becomes clinically and commercially viable. We are aware that*

*NIPT has led to the detection of potential cancer in some pregnant women (Durham C (2015) [Prenatal test detects cancer in mothers](#) Bionews). This is not an issue which we have discussed to date specifically in relation to the detection of cancer and NIPT, but such findings have the potential to raise significant ethical issues and challenges for health professionals. In particular, it raises questions about whether the information should be disclosed when it may or may not be clinically significant and when it could lead, in some cases to overdiagnosis and unnecessary investigations and treatment. In our view, more research is needed to inform discussions and stakeholder guidance to health professionals on this issue.*

The BMA cited their current guidance on the topic which advises that the possibility of other information being identified in testing should be discussed when seeking consent for the test and, if no such conversation has taken place, the presumption should be in favour of disclosing significant information:

*In Medical Ethics Today we provide the following advice in relation to incidental findings and genetic testing in more generally, including how doctors should approach the potential for incidental findings to occur as part of the consent process and on the disclosure of such results, both where this has and has not been discussed prior to testing:*

*“If there is a reasonable chance of other information being inadvertently discovered from a particular test, this should be discussed with the patient...during the consent process in order to ascertain the individual’s wishes about disclosure. The discussion should give examples of the type of information that could be discovered and the procedures that will be followed in that event. When information is discovered unexpectedly, and this discussion has not taken place, the BMA believes there should be a general presumption that significant information will be shared because it would be wrong deliberately to withhold it on the assumption that it would not be in the individual’s interests to know. However, there may be exceptions to this rule, such as where it is judged that revealing the information could cause severe psychological harm to the patient ...When such information is to be given, this must be done sensitively and taking a cue from the individual about how much information he or she is ready and willing to accept at that particular time.”*

The Anscombe Bioethics Centre said that information about the fetus, which was not discussed when the women gave consent to be tested and which indicated the presence of a condition in the fetus for which no treatment is available, should not be disclosed to women.

*Other incidental findings that can arise following NIPT raise very serious concerns. It may be, for example, that a woman tested in relation to a condition that could benefit from early intervention discovers a different condition for which no such intervention is possible, and for which she is offered termination of pregnancy. Incidental findings in relation to the child*

*should certainly not be communicated unless the woman gave permission in advance to the disclosure of such possibilities. In our view, findings where there is no medical benefit for the child or the mother should in any case not be communicated.*

## **5. Who should relay incidental findings**

Felicity Boardman raised an issue concerning who should relay incidental findings, given that prenatal screening professionals may not have the appropriate skillsets for discussing other aspects of the woman's health.

*The health care professionals involved in delivering NIPT are likely not to have expertise in the area of the incidental finding. Who delivers the result, and how any follow-up care is organised might need to be thought through carefully before results are imparted. This might be particularly challenging for the private sector.*

Genetic Alliance UK said that a discussion amongst different kinds of healthcare professionals should take place regarding who should relay incidental findings, in the event that they are disclosed.

*Following the discovery of medical incidental findings, best practice tends to involve convening a multidisciplinary team to discuss the findings and who the most appropriate specialist would be to disclose and discuss these with the woman, so that she is able to make informed decisions about treatment.*

## **6. Implications for other family members**

Felicity Boardman pointed out that incidental findings in genetic testing can reveal information about other members of a woman's family. This creates a question regarding whether this information should be disclosed more broadly, and by whom.

*A genetic abnormality in the mother, discovered as an incidental finding during NIPT, might implicate her biologically related kin. This raises issues about ownership of that incidental finding and who (if anyone) has an obligation to inform family members of the result. This issue might be especially complex if the mother does not consent to her family members being informed.*

**Question 13 - Should potential parents be able to find out the sex of their unborn baby for non-medical reasons from 10 weeks of pregnancy using NIPT? Please give reasons for your answer**

***Potential parents should be able to find out the sex of their unborn baby***

**1. Reproductive choice and autonomy**

It was said by Genetic Alliance UK that it would not be right to stop prospective parents accessing information about fetal sex, acknowledging concerns about sex selective abortion but observing that implementing legal or regulatory rules in this area threatened to undermine women's reproductive autonomy.

*Women should not be prevented from learning the sex of the fetus when receiving NIPT as recommended by the UK NSC....In the UK public opposition to sex selection for non-medical reasons has been repeatedly and clearly demonstrated, including in the form of a specific prohibition in the Human Fertilisation and Embryology Act 2008. However, it is very challenging to enforce this without negatively impacting on the reproductive autonomy of the woman. Any regulation which, for example, attempted to penalise individual women for perceived non-medical sex selection, would not only have very negative consequences for women's rights in the UK, it would also not be effective, as UK regulation is easy to circumvent by travelling or sending a blood sample abroad.*

BPAS also expressed the view that women should be able to access information about fetal sex using NIPT, arguing that concerns about sex selective terminations did not justify restricting women's access to this information.

*The suggestion that women are using information about fetal sex to make decisions about abortion was recently used by MPs opposed to women's reproductive choice to place further restrictions on access to terminations. BPAS worked with organisations supporting women experiencing honour-based violence, and from communities in which son-preference is an issue, to help parliamentarians understand that the solution to issues of gender inequality do not lie in further restricting women's reproductive rights*

Another respondent suggested this was a matter of choice for potential parents and that the information could be useful in enabling certain kinds of practical preparation. Rachel Siden said:

*Yes, with some caution...Revealing the sex before a baby is born is currently routine, and the motivation of most parents is simply to learn this trait out of excitement or to begin purchasing gendered items for their child. Being able to learn the sex earlier does not seem ethically problematic in this context, as the motivations of most parents will remain the same.*

Some respondents who expressed positive attitudes towards the availability of information about fetal sex through NIPT made the point that this was already possible. It was observed by some that women and couples are able to access this information already through other tests. ARC said:

*As women can find out fetal sex from ultrasound scan it doesn't seem appropriate to withhold this information from an NIPT result should they wish to have it.*

It was suggested that it would be difficult to stop women and couples accessing this information. A member of the BMFMS said:

*Yes – this is already available and almost impossible to legislate against. It is better that these tests are available within the NHS rather than driving out a proportion of women into the private sector.*

### **Potential parents should not be able to find out the sex of their unborn baby**

#### **2. Risk of sex selection**

A number of respondents were concerned about the possibility that the availability of this information with NIPT created a risk that potential parents may terminate a pregnancy on the grounds of sex, which would be problematic from an ethical point of view. Another member of the BMFMS observed that because NIPT is available from earlier on in pregnancy this risk was greater than the one posed by information about fetal sex available from ultrasound scanning later in pregnancy:

*I still feel the answer is 'No'. Although it is possible in the UK to organise a social TOP after 15 weeks gestation (when someone might have paid for a sexing scan), this is not easy. Organising a clause C TOP at 10 weeks is much easier. I don't want to live and work in a society where we facilitate TOP based purely on fetal gender.*

The Association of Genetic Nurses and Counsellors agreed.

*It is the view of the AGNC that fetal sexing for non-medical reasons should not be available using NIPT. There would be no medical/economical/disease burden benefits to individuals or society from such testing and it could lead to discrimination on the basis of fetal sex in the decision to continue with the pregnancy*

The CMF argued that accessing information about fetal sex was not objectionable in principle, but could lead to issues in certain contexts.

*That NIPT should provide this information with greater certainty and earlier in pregnancy is not a problem in principle. However, in cultures where there is a bias towards male babies, NIPT may contribute to the pursuit of illegal abortion on the grounds of gendercide.*

Colette Lloyd expressed the view that these kinds of considerations were relevant to policy making in the UK.

*In the mixed culture of the UK, where there are many citizens who would choose to terminate based on gender, (which is currently unacceptable to the GMC), this would increase the risk that this would happen. The NIPT can be done by post, and, as such, the woman could simply have the results in hand and walk into a clinic and ask for a termination.*

The PHG Foundation suggested that there may be cases in which women are put under duress, or coerced by others to terminate a pregnancy on grounds of fetal sex, and that NIPT might be manipulated for these purposes.

*Is there more likelihood of women (especially from some cultures) being forced into early testing and termination of pregnancy? How can women be protected against these pressures?*

Though added that there might also be ethical issues with withholding information that had already been generated from women and couples.

*Alternatively should parents be trusted to have this information at the time it is generated and reported? To withhold information seems paternalistic and inconsistent with a health service which encourages users and consumers to take more responsibility for their health.*

Both DSRF and Saving Down Syndrome cited the PHG Foundation's 2009 report on NIPT which found that the global dimensions NIPT in the context of sex selection might be ethically problematic. DSRF said:

*We agree with the position of the PHG Foundation who stated in their 2009 report [Ethical, legal and social issues arising from cell free-fetal DNA technologies](#), "stressing the advantages of cffDNA technology without paying attention to global dimensions of sex selection might be ethically problematic and likely to weaken public trust in professionals".*

Both BioCentre and the CMF cited an International Bioethics Committee of UNESCO report which raised these issues. BioCentre said:

*Addressing the associated risks of NIPT, a report by the International Bioethics Committee (IBC) of UNESCO stated "another risk lies in the cultural prejudices of preferring a child of the male sex, the sex of the baby being one of the characteristics that can obviously be discovered by NIPT. As this test can be carried out at a very early stage of the pregnancy it would be difficult, even impossible for doctors to forbid the communicating of sex to the parents, and especially at a time when many countries have liberalised abortion. This*

*could lead to a selection based on sex, which is against ethical values of equality and non-discrimination.”<sup>8</sup>*

BioCentre added that there had been accounts of UK doctors facilitating terminations on these grounds.

*In recent years it has been reported that some doctors working in the NHS have been found willing to abort even when there has been no attempt to conceal the fact that the real motivation is gender.<sup>9</sup>*

The Anscombe Bioethics Centre also raised this point and said that there had been no prosecutions for sex selective terminations:

*The termination of pregnancy for reason of the sex of the child compounds the injustice of abortion and should be illegal. Currently the legal status of such abortions is unclear. It is not clear how such procedures fulfil the requirements of the Abortion Act 1967; however, where cases have come to light there have been no prosecutions. Until this legal situation is resolved it would be irresponsible to provide this information, unless in a context where there was no question of termination. As with other information not urgently needed for medical reasons, we do not believe it should be given.*

### **3. No clinical utility**

Some respondents said that this information should not be made available to women and couples since it served no clinical purpose, sometimes mentioning the potential risks. This was one of the points made by the Association of Genetic Nurses and Counsellors, for example, who observed that this information is anyway available to women and couples through ultrasound tests later in gestation, after the point at which abortion is legal.

*Fetal sexing for non-medical purposes remains an option using detailed ultrasound scanning around twenty weeks gestation, when uptake of termination of pregnancy for non-medical reasons is low.*

A member of the BMFMS said that only in situations where there would be benefits to testing for sex-linked conditions should this be offered stating “we should only do additional tests if there is a medical imperative – i.e. before invasive testing for sex linked conditions” and this was also the view of Clinical Genetics and Cytogenetics, Guy’s Hospital.

*No. Finding out the sex through NIPT at 10 weeks should only be offered for at risk patients (with a family history of X-linked disorder for example). This may increase again TOPs for wrong reasons and should be avoided.*

---

<sup>8</sup> See BioCentre’s full consultation response for references.

<sup>9</sup> See BioCentre’s full consultation response for references.

#### 4. Poor use of NHS funds

Some said that making NIPT for fetal sex available on the NHS, specifically, would not be a good use of state resources. Lorna Watson said this was especially problematic given the risks described above relating to selective termination on grounds of sex.

*This would be an inappropriate use of NHS resources and would be likely to lead to sex selection abortion which is unlawful, even if it is not declared with this intention.*

The PHG Foundation, who argued that prospective parents should in principle be able to access this information, agreed that such access should not be publically funded.

*In a hypothetical situation where the woman has declined NIPT for any medical reasons but wants to have NIPT purely for information to learn the fetus' sex, while this would likely be acceptable ethically, it would also be reasonable for the NHS to decline to fund this, as in an environment of very limited resources this funding could achieve greater patient benefits invested elsewhere in the health and care system.*

**Question 14 - What genetic information, if any, do you think parents should be allowed to find out about their unborn baby using NIPT? Please give reasons for your answer.**

**1. All information**

Some respondents said there was no information that women should not be able to access and that emphasis should be placed on quality counselling and information. ARC said that parents were best placed to make decisions about what information to access and expressed the view that concerns about people using NIPT to produce designer babies were misplaced.

*The important point is that parents understand the limitations, potential harms and benefits of any test before deciding to have it. With the prerequisite of accurate pre-test information and counselling, parents are best placed to decide on their information needs and preferences. From our long experience at ARC of contact with parents and professionals we do not share the fear that parents seek perfection or a 'designer baby'.*

This was the view of BPAS which also thought that women should be able to access what was essentially patient information to which they were entitled.

*As in all areas of healthcare there is a patient, and in this particular context the patient is the pregnant woman...It is vital that healthcare professionals are not led to believe that pregnant women are not patients in their own right, but simply mothers-in-waiting and just one part of a couple. With that in mind any information that can be gathered about a patient that the patient wishes to know should be given to them.*

**2. Chromosomal disorders**

Some respondents commented on the current state of the art in NIPT and expressed views on what they thought NIPT could and should be used to test for at the moment. A member of the BMFMS said that, currently, evidence for the accuracy of NIPT in areas other than Down's, Edwards' and Patau's syndromes and sex testing was not good enough, and that information relating to other conditions or traits should not be accessed.

*At this stage I would suggest T13, 18, 21 and gender. There is limited data on other tests in terms of accuracy. It is important to establish governance and quality control in line with current choices (combined screening screens for 13, 18, and 21 and gender determination is routinely offered at 20 week scan) before considering extending the scope of screening tests offered.*

### 3. Medical conditions

Some respondents said that information relating to health conditions should be available to women and couples. Felicity Boardman, for example, said:

*Information relevant to the health and mental well-being of that future child.*

Genetic Alliance UK agreed, pointing to the World Health Organization guidance on prenatal diagnosis as a potential guide to good practice in NIPT.

*The World Health Organization's Proposed Ethical Guidelines for Prenatal Diagnosis apply here, with the key principle being that prenatal diagnosis should be performed only for reasons relevant to the health of the fetus and only to detect genetic conditions or fetal malformations.*

### 4. Serious medical conditions

Some defined more narrowly the group of conditions about which they thought women and couples should be able to access information using NIPT, and said explicitly that these should be severe, important or urgent medical conditions. Clinical Genetics and Cytogenetics, Guy's Hospital said that information about "early onset severe disorders" should be accessible to women and couples and an anonymous respondent said "only medical conditions that lead to death or a significant risk of severe handicap." Lorna Watson said:

*The information should be limited to important medical conditions where there is benefit to be conferred from the knowledge in antenatal care for the mother and wellbeing of the foetus. This should not include those conditions compatible with good quality of life where terminations could increasingly be seen to be an option by some.*

The Anscombe Bioethics Centre said NIPT should be available where information was needed urgently for medical reasons.

*NIPT should not be routinely provided, but at most, provided to parents who have an urgent medical need to know e.g. to protect their child's health so that prenatal treatment can be given.*

Though they added that it should be made clear to women and couples in cases where termination was the only available intervention that this was the case, adding:

*Parents should be given every opportunity not to be placed in a situation in the future where they may face pressure to abort their child.*

### 5. Treatable medical conditions

Some respondents thought that the category of conditions for which women and couples should be able to access information with NIPT was narrower, and said that

only information about conditions for which medical treatments were available should be accessible. Saving Down Syndrome said:

*The only genetic information which can be of any real use for parents is that which can lead to therapeutic (life affirming) intervention, at the optimum time.*

And the CMF agreed that only in cases where therapies were available for the tested for conditions should this information be accessible to women and couples.

*Whatever information, genetic or otherwise, that NIPT brings to light, should be used only to enable medical staff, parents and other family members to be better prepared for the arrival of the new baby or to enable therapeutic interventions (medical or surgical) to improve the baby's health before birth or in the perinatal period.*

## **6. Societal implications**

Alongside comments about particular kinds of information that should be accessible to women and couples, some respondents also raised concerns about the long term effects on society of adopting liberal policies on the use of NIPT. DSRF UK said:

*Under the current climate of disability discrimination within screening we cannot see it panning out well for society. People will demand their unborn child's DNA as it is perceived as their property, to do with as they will. We do not see how this will benefit them in most cases and, as someone (Dr Corcoran) who has seen her own DNA results, they are confusing and hard to interpret. Every person will have conditions and diseases they will be at risk of. We do not understand the illnesses/conditions and their environmental interplay enough to deliver life estimates (which will be asked for).*

A member of the BMFMS also said that issues about how women and couples should be able to make use of NIPT went beyond those relating just to individual rights:

*Although this is mostly about consent and pre test counselling, I do think that this is a slippery slope... where to draw the line? And this line will change anyway as we learn more and more. I think a line does have to be drawn. Should the 'rights to all knowledge' of an individual be prioritised over what might be better for society in the long run?*

## **7. NHS funded services**

Another point made concerned specifically the use of state funds and what should be available as part of NHS antenatal care. Clinical Genetics and Cytogenetics, Guy's Hospital said that:

*If the NIPT is provided by the NHS then only clinically relevant and reliable information fulfils the UK NSC criteria, including early onset severe disorders and actionable late onset disorders.*

## **8. Relevance of existing guidance**

The Association of Genetic Nurses and Counsellors said that the approach used in invasive prenatal diagnosis could be used as a model to guide policy or clinical decisions about the appropriate uses of NIPT.

*It could be argued that parents should be able to find out the same sort of information about their unborn baby through NIPT as for invasive pre-natal diagnosis as the ethical framework for the latter has been long established in Clinical Genetics and Fetal Medicine. It would be important for couples to be counselled appropriately beforehand about why they want this information, to reduce the likelihood of babies being born who have already been tested for adult-onset conditions, for instance.*

**Question 15 - What genetic information, if any, do you think parents should not be allowed to find out about their unborn baby using NIPT? Please give reasons for your answer.**

**1. None**

Some respondents said or suggested that all information should be made available to women and there should be no restrictions. ARC said:

*On the whole, it is our experience at ARC that parents are able to make responsible decisions about testing... We at ARC have a firm belief in reproductive autonomy and would not want to see this compromised.*

BPAS expressed a similar view.

*[Healthcare professionals] must trust all patients, including pregnant women, with information about their health, which they can then use to make the right decision for them.*

The BMA also said that information should not be restricted to, for example, cases in which women might be considering terminating their pregnancy. They added, with the caveat that NHS resources should be spent proportionately, that women should not be expected to commit, before prenatal screening, to a decision about terminating a pregnancy in light of information yielded by NIPT.

*Restricting access to women who would consider terminating a pregnancy would be, in our view, inappropriate. As stated above, as much information as possible should be given to women and their partners to make an informed decision and whether to opt for testing and if so how to respond to an unfavourable result. Although it is important to use limited resources carefully in a publically-funded health system, "clinical benefit" is much broader than pregnancy management and it is unreasonable to expect women to decide in advance how she would respond to an unfavourable result.*

**2. Information not related to serious medical conditions**

Some respondents said that only information about serious, important or severe conditions should be communicated. An anonymous respondent said that information about " *medical conditions that are easily treated or do not lead to death or a significant risk of severe handicap*" should be not be accessible and Lorna Watson said that information about conditions that are compatible with high levels of wellbeing should not be accessed.

*The information should be limited to important medical conditions where there is benefit to be conferred from the knowledge in antenatal care for the mother*

*and wellbeing of the foetus. This should not include those conditions compatible with good quality of life where terminations could increasingly be seen to be an option by some.*

### **3. Information that cannot be used for prenatal treatment or preparation**

Some said that it was important that information accessed through NIPT could inform decisions about possible treatment or alternative means of preparation. DSRF UK said "...at present the only genetic information that will provide benefit is that which will allow access to time sensitive treatments or health improving interventions" and the CMF stated that:

*Whatever information, genetic or otherwise, that NIPT brings to light, should be used only to enable medical staff, parents and other family members to be better prepared for the arrival of the new baby or to enable therapeutic interventions (medical or surgical) to improve the baby's health before birth or in the perinatal period.*

A member of the BMFMS suggested that only information that could inform decisions about either interventions or terminations should be available. They said that information should not be accessible that could:

*...a) not lead to termination under clause E (either because there is no parental intention to do so or because the condition was deemed insufficiently serious to justify) b) not result in any treatment for the neonate that had a chance of improving outcome e.g. yes test for CAH to determine sex and as steroids might be started early but no to test for BRCA.*

Saving Down Syndrome said that information that could be used to make 'life affirming' interventions only should be available.

*The only genetic information which can be of any real use for parents is that which can lead to therapeutic (life affirming) intervention, at the optimum time.*

The Anscombe Bioethics Centre said that such interventions should be urgent.

*Information not urgently needed for a genuinely therapeutic purpose such as prenatal treatment should not be communicated. The child in utero is in a very vulnerable situation with very limited legal protection and dependent on the attitudes of others for the character of his or her welcome into the world.*

The Liminal Spaces Project also warned about the risks of testing for information beyond medical necessity and highlighted the issues that might be raised for the person the fetus might later become.

*It is important to highlight to all concerned that there is a possibility that testing beyond immediate medical need for the fetus - such as whole genome sequencing - might lead to a closing down of options for the potential future person rather than an opening up of options. Consider, for example, the insurance implications for the future person of having information permanently on their record. She/he will be deprived of the option of not seeking or knowing certain health-related information about him/herself. The nature of the insurance contract is such that all known information potentially pertinent to a policy must be disclosed (it is a contract of the utmost good faith).*

#### **4. Less certain information**

Some concerns about information yielded by NIPT related to the confidence in which women and couples were able to have in its accuracy or implications. One reason for this related to the accuracy of NIPT in identifying different kinds of information and the predictive values, sensitivity and specificity of the test for different conditions. A member of the BMFMS said the accuracy of NIPT in identifying a given piece of information was an important consideration in whether it should be accessible to women and couples.

*At this stage all tests other than above until robust data is available about accuracy of other tests.*

A different kind of concern related to our understanding of genetics itself and how well we are able to interpret the implications for individuals of the genetic information that NIPT can identify. The PHG Foundation said that most information accessible with NIPT should not be conveyed to women and couples as we do not currently have a good understanding of its implications.

*...The majority of genetic information should not be communicated. This would be on the basis that we are at a very early stage of understanding about the significance of genetic information and the extent to which this is predictive of future disease. Thus our knowledge is very preliminary: we need to understand far more about the penetrance of disease; the extent to which different types of risk factors combine; and the incidence of diseases (even in diseases like breast cancer caused by the BRCA1 and 2 variants, which have been well-described, we have limited understanding of this disease in families without a family history of disease).*

#### **5. Non-medical information**

Some concerns related to information not related to health. It was said that information relating to some non-medical features should not be relayed to women and couples. The Association of Genetic Nurses and Counsellors said:

*Parents should not be allowed to find out non-medical information that would not be relevant to the child's health, such as information about traits. This*

*could lead to discrimination on the basis of non-medical traits, such as appearance or intelligence, in the decision to continue with a pregnancy.*

Felicity Boardman agreed that only health related information should be accessible.

*I don't think that parents require access to any information that does not relate to the future health and mental well-being of the child. I would include within this information on characteristics such as hair and eye colour, intelligence and any other characteristic that is not connected to health issue.*

Genetic Alliance UK said that good practice guidance should be used to support healthcare professionals to discourage women and couples from accessing information about non-medical information as well as the presence of recessive genes.

*For similar reasons, and to the same degree, that parents are discouraged from testing their children for late onset genetic conditions, women should be discouraged from accessing nonmedical information about their fetus. This point applies too to nonpertinent medical information, such as carrier status. This is an issue best addressed, through ethical guidelines and best practice tools, rather than legislation or regulation.*

Kay Sammon expressed worries about the implications of use of NIPT to access information about non-medical features, given that law and regulation varies in different parts of the world.

*A future of 'designer babies' is of great concern particularly without regulation across borders.*

## **6. Late onset conditions**

A number of respondents raised issues concerning the use of NIPT to find out about late onset conditions, such as Huntington's disease, which develops in middle age. Clinical Genetics and Cytogenetics, Guy's Hospital thought that information about late onset conditions, for which there are no treatments, should not be conveyed to women and couples.

*For NHS funded testing, anything that is not clinically relevant and results that indicate non-actionable late onset disorders.*

Colette Lloyd agreed that information about such conditions should not be relayed to women and couples, since the decision to access this information should be left to the person the fetus becomes.

*...Genetic testing should not be allowed for conditions that won't develop until adulthood – the decision to test for these can be made by the adult themselves.”*

Though ARC were broadly supportive of women and couples having access to all information they agreed that late onset conditions might be an area where difficulties could arise.

*There are challenging issues surrounding late onset disorders which may lead to an encroachment on the child's right not to know. But we would not want to see an overly paternalistic approach to the information that is or isn't provided to parents. We at ARC have a firm belief in reproductive autonomy and would not want to see this compromised.*

## **7. Spectrum disorders**

Colette Lloyd agreed that only information that could support decisions about prenatal treatment should be conveyed said that there were issues with making accessible information about conditions that give rise to symptoms or features of a range of levels of severity.

*It should be for genetic conditions where the outcome is nearly identical in all cases, and not affected by other things. In many conditions, such as Down's Syndrome, just because the genetic make-up is known, it does not dictate what the outcome for that particular person will be.*

**Question 16 - Do you think whole genome sequencing of unborn babies using NIPT should be allowed? Please give reasons for your answer.**

***Whole genome sequencing of unborn babies using NIPT should be allowed***

**1. Whole genome sequencing as means of testing for genetic conditions**

The BMA observed that since whole genome sequencing was able to reveal information about a range of genetic conditions it could act as a 'universal' test for genetic diseases for which there are no other tests, and said that making this information available to women would have benefits.

*The current applications of NIPT are restricted to fetal sex determination, some single gene disorders which are inherited in a dominant fashion from the father or which arise de novo, and for aneuploidies. By contrast, sequencing the full fetal genome has the potential to detect any of the single gene disorders known to exist, genetic mutations associated with other conditions and, in principle, de novo mutations. [Non-invasive fetal whole genome sequencing] therefore might act as a universal, non-invasive prenatal test for all diseases or conditions which have a genetic cause or component. It could provide a significant benefit to pregnant women at risk for genetic conditions but who are only currently offered invasive prenatal testing and it may also help to determine previously unexplainable fetal abnormalities or losses. If no other, more directed, tests exist, there is a clinical benefit to testing via [non-invasive fetal whole genome sequencing].*

The PHG Foundation, who were broadly critical of many uses of whole genome sequencing, made a similar point suggesting that whole genome sequencing technology might be used to test for variations for which there were already tests, as long as the performance of NIPT for identifying such variations had been established.

*...It is possible that targeted WGS of unborn babies using NIPT as a 'virtual panel test' could feasibly be used as a replacement technology to detect genomic aberrations that are already tested for or where tests are currently in development. Using combined WGS and NIPT in this way would require sufficient technical equivalence, sensitivity and specificity to be achieved.*

## ***Whole genome sequencing of unborn babies using NIPT should not be allowed***

### **2. Inadequate levels of genetic understanding currently**

As with responses to question 15, some respondents indicated that they thought that our current levels of understanding of human genetics were not good enough to make collecting the amount of information whole genome sequencing would enable, a worthwhile, or wise, enterprise. One member of the BMFMS said that whole genome sequencing should not be accessible at the moment since there is “...*too much we don't know or understand. Too many uncertainties...*” and another BMFMS member said that such uses could only be supported in cases where there was evidence of the technical strengths of such testing.

*Only when / if there are robust publications as to the significance of VOUS and real systems in place to monitor both sides of the screening equation sensitivity and specificity.*

The PHG Foundation concurred.

*The PHG Foundation opposes offering whole genome sequencing of the entire fetal genome of unborn babies using NIPT on a universal basis. It is premature to offer such testing within routine clinical care or public health screening, on the basis that there is not sufficient understanding of what the results of WGS testing means to be able to interpret results reliably, and understand the clinical meaning of the results for the unborn child and their family. In particular, the penetrance of many genetic diseases is not sufficiently understood.*

The wide range of hard-to-interpret information that could be generated by whole genome sequencing would raise issues for healthcare professionals seeking consent from women to undergo testing, the BMA said:

*As with all genetic testing, informed consent would be important and at the outset any consent process should include information on the range of information that would be produced and the difficulties this could raise. However, given the range of information which NIFWGS could yield and the potential for there to be detrimental, but as yet unknown, consequences for the interests of the future child, obtaining and being assured that a patient has provided informed consent would be difficult.*

### **3. No clinical utility**

Some concerns were raised about enabling women and couples to access information about the fetal genome relating to the purpose or use to which that

information could be put. A BMFMS member suggested that there would be no utility in gathering this data: “*Not at the moment – what would we do with that information...?*”

The BMA agreed that ethical issues were raised by seeking information about conditions for which there were no available treatment options, or seeking information purely for its own sake.

*Where [non-invasive fetal whole genome sequencing] is sought just for information purposes, or where the information that is disclosed to women and their partners will not result in direct clinical benefit, this does have the potential to cause ethical tensions. Whole genome sequencing could potentially provide parents with information about their fetus in relation to all diseases or conditions with a known genetic cause or component. This is irrespective of the seriousness of the disease, the onset of the condition, the level of risk, or whether the provision of information would result in some benefit. [Non-invasive fetal whole genome sequencing] could also provide prospective parents with findings of unknown clinical utility and about nonmedical inherited traits.*

Some respondents saw the ethics of this situation similarly to others in which information from NIPT could not inform decisions about medical treatment and cited broader principles about the need for the availability of therapies to justify accessing the information. Saving Down Syndrome made this point.

*The only genetic information which can be of any real use for parents is that which can lead to therapeutic (life affirming) intervention, at the optimum time.*

#### **4. Implications for the future child**

Some raised concerns about the impact on the future child and adult that the fetus might become, if women and couples were able to access all available information about their genome. The BMA cited good practice in the genetic testing of children in relation to this issue.

*If the decision is taken to carry the pregnancy to term, a key issue is the impact that [non-invasive fetal whole genome sequencing] may have on the autonomy and privacy interests of the future child. Currently, in relation to the genetic testing of children, parental access to information which would not provide some tangible benefit to the child is either restricted or at least discouraged, on the basis that a child’s genetic status is his or her own private information and intervening into this private sphere without justification would be inappropriate. [Non-invasive fetal whole genome sequencing] could mean that this information is generated and potentially disclosed prenatally. It may include information which is relevant to adult onset conditions, for which*

*testing would usually only be undertaken once the person could consent to testing, or in relation to a susceptibility to disease, for which testing would also not typically be conducted on a child.*

This might be problematic, the BMA said, for reasons relating to individual choice about accessing information about one's genetics, or practically, in terms of, for instance, accessing insurance.

*Although legally the disclosure of this type of information to a woman about her fetus may not be problematic, it does have the potential to raise significant ethical issues. As individuals cannot “unknow” what may have been revealed through [Non-invasive fetal whole genome sequencing], its disclosure and retention may restrict their ability to make choices about their health – including the choice of whether or not to find out information about their genetic make-up. It could also have other negative practical implications, for example when applying for insurance in the future. This is particularly important given that it is uncertain how genetic information may be used in the future.*

Similar general principles underlying good practice in genetic testing of children were invoked by Genetic Alliance UK, who said that only in situations in which medical options were immediately available should prenatal whole genome sequencing be available.

*There are concerns relating to the potential impact on the autonomy of the future child if whole genome sequencing using NIPT were permitted. These ethical issues are very similar to those which arise in the related situation of genetic testing of non-competent children, and in our opinion very similar principles apply. Genetic testing of children should generally only be considered acceptable where treatment or surveillance will be instituted or altered. Where the test will not be of medical benefit to the child for some time, there should be a presumption of delay until a child is old enough to choose for him or herself, unless there are compelling reasons to test earlier.*

However, they said that the information about adult onset conditions provided by NIPT could be of relevance to decision making in pregnancy, given the availability of abortion.

*For prenatal testing this question is complicated by the possibility of termination of pregnancy, an option not available postnatally. Thus while predictive testing is regarded as unacceptable in childhood, it can be of relevance to the reproductive autonomy of the woman, even if relating to an adult onset condition for which there is no treatment. Studies have shown that it is very difficult to predict a women's decision making about whether to continue a pregnancy and why, even if she has quite strong views beforehand.*

## 5. Selective termination

Worries were raised about women and couples using information gained from whole genome sequencing to inform decisions about terminating pregnancies.

*If combined WGS/NIPT testing were used in this way the provision of WGS in unborn babies might result in increased numbers of terminations of pregnancy or could result in overdiagnosis and medicalisation.*

Lorna Watson said:

*This may give rise to concerns about conditions which may not manifest in the phenotype, and increase anxiety, and lead to genetic selection of the population in the longer term. Full public debate is needed on this issue.*

## 6. Storage of information

Issues relating to the storage of genetic information generated by prenatal whole genome sequencing with NIPT were raised by some respondents. The BMA explained that this was an issue that they had raised with the Information Commissioner.

*There are also further questions relating to whether, and if so under what circumstances, retention of the information generated from NIFWGS would be permitted under current legislation. We have had some initial discussions with the Information Commissioner's Office about this and have also begun to consider how, and to what extent, information derived from such testing would be stored and communicated as the child gains competence to make decisions. This is an issue that requires further discussion.*

Genetic Alliance UK also raised this issue and agreed that that there should be public debate about what the appropriate protocols for treatment of prenatally collected genetic information might be.

*We feel that this is an area where a broad public conversation may be required, in order to find general social values which can be applied. For example, it may be considered appropriate by the majority of the population to store WGS data unanalysed, except for examination of information of immediate medical relevance to the potential child. This then allows the child the option of learning more about their genetic material, when they reach the age of competence to make that decision.*

The PHG Foundation pointed out the opportunities in this context. They said that data should be collected on pregnancies analysed with NIPT to create an evidence base on which future whole genome sequencing-based prenatal testing might be founded.

*If population wide NIPT is implemented for aneuploidy screening, it is vital that anonymous test results are able to be linked to a national register of pre-natal, birth and extended post-natal follow-up. A comprehensive programme of data collection, registration and evaluation of all pregnancies analysed by NIPT is needed in order to construct the evidence base on which future WGS-based prenatal testing could accurately be provided.*

## **7. Societal implications and eugenics**

Some expressed the above point more strongly. One anonymous respondent said unless whole genome sequencing with NIPT was used as a means of identifying serious conditions that were not treatable, this raised concerns about eugenics.

*No – leads to eugenics. A wider panel of targeted tests for untreatable severe conditions might be acceptable. The benefits of early detection of less severe or modifiable diseases can be achieved through postnatal testing.*

A related concern was that people may make use of NIPT to enable them to have ‘designer babies’ and this may impact on diversity and tolerance in society.

*A future of ‘designer babies’ is of great concern particularly without regulation across borders. Will we live in a world where certain people are ‘screened out’? What effect will this have on diversity and tolerance? Darrin and Dixon (2009) concluded that although progress has been made through improved legal rights and policy change, in reality considerable inequalities still exist today as a result of societal prejudices and negative attitudes towards disabled people, with societal pressure to have a ‘normal child’.*

**Question 17 - What, if anything, might the increasing availability and use of NIPT mean for people living with genetic conditions? Please provide evidence or examples if possible.**

**1. Less investment in research, services and support**

Some respondents worried that there may be implications for the funding of research into genetic conditions. DSRF UK were concerned that “*research will be stifled...*” and Lorna Watson made a similar point, expressing the view that funding directed towards research relevant to conditions for which screening is possible is disproportionately weighted towards advancing screening technology.

*It would result in a reduction in research funding for conditions, already heavily imbalanced for towards screening and termination rather than research supporting people with the condition.*

The PHG Foundation were concerned that women and couples who chose not to screen or to terminate may be viewed with disapproval by the state and others, which may result in reduced access to state support and services.

*There is also a fear that families who chose to proceed with a pregnancy knowing that they will have, or are at risk of having a child affected with a genetic condition, will be regarded as ‘irresponsible’. This could result in the increased financial burden on these families for care of children being regarded as legitimate and their responsibility, and hence not eligible for wider state support.*

**2. Reduction of number of people with Down’s syndrome and its implications**

Some concerns related to the potential impact of NIPT on the number of people with Down’s syndrome in the UK. Some respondents suggested that the prevalence of the condition may decline and cited issues for people with Down’s syndrome, were that to happen. Some thought that this outcome may bring about, or worsen, some of the potential implications already mentioned. Colette Lloyd, for instance said that a reduction in the prevalence of the condition would mean that health, social and education services for people with Down’s syndrome may be reduced.

*...A reduction in the number of people with Down syndrome may also lead to diminished structures and resources for their healthcare, well-being and education.*

The Anscombe Bioethics Centre were also concerned about potential impacts on the availability of support for and on knowledge of the condition.

*There will be fewer of their peers surviving with Down Syndrome, which will also adversely affect them as their parents will receive less support from other parents of Down's children and there will be less understanding of their condition. While of course it would be wrong deliberately to cause Down Syndrome or other genetic conditions by some hypothetical genetic intervention, simply allowing those with Down Syndrome to be born like any other child is not 'causing' the condition but simply respecting the individual concerned.*

They also cited the impact of there being fewer other people with Down's syndrome on the experience of having Down's syndrome and suggested that opportunities for bonding and solidarity between those with the condition would be diminished.

*The solidarity resulting from the survival and bonding of children and adults with Down Syndrome and other genetic conditions is a very good thing, and its absence will mean that parents who do have surviving children are more likely to be left in a lonely, unsupported and ill-informed state in their local community. Such bonding is infinitely better than the absence of children who are no longer present in the community simply because the community has taken it upon itself to exclude them definitively at their parents' request on grounds of their disability.*

Felicity Boardman agreed that there could be benefits for people with a given rare condition of knowing and interacting with others with the same condition, citing her own work with people with Spinal Muscular Atrophy.

*My research has shown that for many people with rare conditions, meeting others with the same condition, and indeed, seeing others thrive with the same condition is both an affirming and validating experience. On a human level, we all want to see our lives and experiences reflected and represented in the world around us- whether this be through the media, or through the people around us. Having other people who look, or experience the same physical differences as us is important, and people with disabilities are a key source of support for one another. This sense of support and community would be threatened by screening designed to reduce the number of people with those conditions coming into the world.*

Felicity Boardman also thought that lower numbers of people with a given condition would create issues for those seeking research funding into those conditions and pointed out that, given that resources for research funding are limited, it can be harder to justify decisions to fund research into less common conditions.

*As the number of people living with these conditions declines, so too would the justification of funding budgets for research into treatments and cures.*

Others were less confident that a reduction in the number of those with Down's syndrome would be the outcome of widening availability of NIPT. The DSA said that this was an open question but suggested that this matter should be kept under review.

*It is not yet clear what impact, if any, the increasing availability of NIPT will have upon the number of people born with Down's syndrome in the future. The issue will need to be closely monitored.*

And Genetic Alliance UK, conceding that this may be one result of broader access to NIPT, said that even if this were one outcome of widening use of NIPT, it would not necessarily raise ethical issues different to those raised by use of vaccines and other medical interventions aiming to eliminate serious disease.

*While it is true that widespread termination of affected pregnancies is likely to reduce the number of affected children born with a specific condition, which would cause the condition to become rarer, with all the resulting implications for research funding, medical expertise etc., this is not substantially different in effect from the impact of vaccines and other approaches to the eradication of disease.*

### **3. Devaluing of disabled lives**

A number of respondents were concerned that expanding prenatal screening programmes with NIPT may devalue disabled people. Colette Lloyd said that “*there may be a sense of lower value inflicted on the people who have the genetic condition, caused by a presumption that is prevalent in society today that a responsible citizen would terminate a baby with such a condition and try again*” and Kay Sammon suggested that it raised questions about the need for:

*...A broader ethical debate about the value (or lack of) placed upon the lives of disabled people when in many other aspects of society there is more inclusion than ever before.*

The Anscombe Bioethics Centre said that this may have quite direct consequences in terms of how people with genetic conditions are affected, and could manifest as offence taken, or hurt experienced.

*Adults with Down Syndrome, for example, are often quite able to understand the meaning of prenatal tests for identifying Down's: rightly, they will judge that they are not sufficiently valued by a society which offers such tests in practice specifically to exclude them.*

A member of the BMFMS was also concerned about this outcome.

*A better level of understanding in the population of genetic disease may be a good thing for those affected by these conditions, but the offer of prenatal testing (and therefore termination of pregnancy) may serve to devalue these individuals.*

BioCentre expressed concerns that prenatal screening conveyed a message that the lives of disabled people were of less value and cited disability charity Disability Awareness in Action.

*These tests foster the notion that the lives of people with disability are ‘not worth living’ and that it is socially desirable to prevent people with disabilities from being born<sup>10</sup>. As Dr. Rachel Hurst, Director of Disability Awareness in Action, notes “There is little doubt that the modern world is not a good place for disabled people to be born into”<sup>11</sup>.*

Felicity Boardman expressed similar concerns.

*People with the screened-for conditions (and other disabilities) may feel that the existence of the screening programme (underscored by the introduction of NIPT) sends a negative message about the value of the lives of people with disabilities. The screening programme might be interpreted by these people as sending the message that society would have preferred it had they not been born.*

However one anonymous respondent said that arguments that prenatal screening devalues the lives of disabled people involve a confusion.

*When dealing with a disabled child, it seems that many campaigners find it hard to distinguish between the child and the condition. This is not something I have ever struggled with. My daughter is a beautiful child. She is a full member of our family, she has her own likes and dislikes, she has an enchanting smile and a wicked laugh, she loves food, especially lamb tagine, and like all girls, she wears an unnecessary amount of pink clothing. I love my daughter. I hate lissencephaly...Lissencephaly stops her from being everything that she could have been, everything that she should have been. I do not accept this state of affairs as being somehow “as good as” it could have been had she been born without lissencephaly. Not having Down’s is better than having Down’s. Being able to see is better than being blind. Being able to walk is better than not being able to walk. There is no moral equivalence, and there should be no equivocation about this.*

#### **4. Discrimination, isolation and violation of rights**

A number of points raised by respondents concerned the possible implications of growing use of NIPT for how people with genetic conditions are seen, and treated, by wider society. Colette Lloyd was concerned that increased use of NIPT could “increase stigma, reduce public inclusion and tolerance”. Some respondents thought that widening access to NIPT raised questions about disabled peoples’ rights and raised the threat of discrimination. The CMF cited the campaigning organisation Don’t Screen Us Out.

*To quote from the Don’t Screen Us Out campaign: ‘To introduce a screening test that would enable the increased selective elimination of children with*

---

<sup>10</sup>See BioCentre’s full consultation response for references.

<sup>11</sup>See BioCentre’s full consultation response for references.

*Down Syndrome due to a lack of proper inclusion, accommodation, and support, would violate disabled rights and our ethical obligations to disabled people and communities.'*

The PHG Foundation said that this stigma and discrimination might be felt both by disabled people and their families.

*... There are fears that the increasing availability and use of NIPT might result in greater stigmatisation and discrimination against individuals and families with genetic conditions.*

However, ARC resisted the idea that improving screening options would be bad for people with genetic conditions.

*Screening for genetic conditions has been available for three decades and this has coincided with real gains by disability rights advocates. While there is still much work to be done in creating an inclusive society, raising awareness and particularly around ensuring there are adequate resources and support available to those living with genetic conditions, it is our belief that giving parents the choice to have NIPT will not be to the detriment of those living with genetic impairments. We know that many of those living with impairment and their advocates share our thinking – restricting choice for prospective parents is not the way to achieve positive benefit for their community.*

And CARE suggested that they saw the situation as analogous to ones in which eugenic social policies had been adopted.

*Despite the best of intentions, the introduction of NIPT on the NHS would inevitably have the effect of introducing a new crudely utilitarian ethic into British society with some disturbing parallels to that embraced in Nazi Germany which would fundamentally threaten to compromise the compassionate nature of our society.*

## **5. Benefits of reproductive choice**

Some respondents emphasised the potential for enhanced reproductive autonomy that NIPT could provide people with genetic conditions. Clinical Genetics and Cytogenetics, Guy's Hospital proposed that there might be a range of potential benefits.

*NIPT for people living with genetic conditions would allow early prenatal diagnosis when the transmission risk is high (i.e. autosomal dominant conditions). It would allow earlier TOPs if the fetus was affected. It would also allow avoiding invasive testing and its associated complications.*

ARC agreed, stressing that an option for undergoing testing without needing to have an invasive procedure would be welcome for some women.

*When it comes to NIPD, for some parents who have or carry genetic conditions it could be of benefit if they wish to be informed as to whether their baby might be affected without undergoing invasive procedures.*

Genetic Alliance UK made the same point, adding that the information yielded by NIPT may be of use to people with genetic conditions, whether or not they anticipated choosing to terminate a pregnancy.

*... For people at risk of some genetic conditions there is value in prenatal diagnosis even if they have not made the decision to terminate an affected pregnancy. For example, prenatal diagnosis of congenital adrenal hyperplasia permits early management of the pregnancy to prevent virilisation, which can significantly reduce the impact of the condition on affected individuals" Similarly, in Rhesus negative mothers early knowledge of the fetal blood type can avoid unnecessary monitoring and prophylaxis.<sup>12</sup>*

---

<sup>12</sup> See Genetic Alliance UK's full consultation response for references.

## Question 18 - Is current regulation covering the provision and marketing of NIPT in the UK sufficient and appropriate?

### 1. Role of professional bodies

Some respondents said that further regulation of the provision and marketing of NIPT was not needed but suggested that using alternative means of ensuring responsible provision of NIPT might be appropriate. The Liminal Spaces Project, for instance, expressed the view that there was no need for additional legal instruments but said that professional organisations had responsibilities in this area with respect to training and guidance.

*We do not believe that more hard law regulation is required in this area. Rather, we suggest that attention is paid to exploring and understanding the ethical issues and responsibilities at stake. The role of professional bodies becomes crucial here, not just in ensuring that their existing guidance is up-to-date, but also in making available appropriate training for their professional members both in the technicalities and limits of this technology, and in helping professionals to work with and for patients and citizens who seek out this form of testing. This extends both to tackling the nature and scope of informedness when it comes to obtaining valid consent, and to the underlying tensions that exist when the availability of this kind of testing is simplistically presented merely as a matter of choice. The framing of the issues and the potential consequences appropriately – for and with patients and citizens – is of crucial importance.*

ARC observed that the Care Quality Commission already had powers to tackle the worst practice, though acknowledged that there were challenges in encouraging, or enforcing, high standards. They also suggested that there was a role for professional bodies, including medical royal colleges, to provide advice and guidance.

*While the CQC and other bodies can prevent very bad or unsafe practice, there is much that goes on in the private sector provision of NIPT that is far from optimal. However, it has to be said that this is also the case for many other areas of private health care. It is hard to envisage where more effective regulation will come from and whether there is actually any appetite for it among policy makers who remain in thrall to the market. Pragmatically, we would look to professional bodies, including the royal colleges to produce expert evidence-based advice and good practice guidelines for NIPT providers. This may go some way to exposing outliers. Furthermore, expectant parents could be encouraged to check that the clinics they are considering can show that they adhere to such published guidance.*

The DSA observed that existing NICE guidance covering the provision of patient information and support was of relevance to the delivery of NIPT but stated that this was not being followed.

*...NICE guidelines are not being met in relation to the provision and delivery of information. Non-compliance with NICE guidance is having a negative impact upon parental experiences of the screening process and their ability to make an informed choice about the future of their pregnancy.*

## **2. General concerns about regulation**

Some non-specific concerns were expressed about the regulation of NIPT. A member of the BMFMS responded to this question “*probably not*” and another BMFMS member said “...*my gut feeling is ‘no’.*” Some particular areas were highlighted by respondents, as below.

## **3. Regulation of information and marketing is inadequate**

Some respondents raised issues relating to the marketing and promotion of NIPT by private providers and concerns were expressed about how NIPT is described by private providers. The Anscombe Bioethics Centre cited both marketing and provision as areas in need of further regulation.

*No. There should be very strict restrictions on provision and marketing of NIPT in the current climate, where negativity and misinformation about the medical conditions tested for is unfortunately rife.*

Felicity Boardman agreed that regulation is inadequate currently and that information provided by private clinics was sometimes poor, highlighting the conflict of interests experienced by private providers, given their objectives to sell tests.

*No- within the private sector NIPT is not regulated. Companies provide inaccurate information about the screened-for conditions in order to sell screens.*

The BMA said that whilst regulation in this area could be tightened in the UK this may not be sufficient to address concerns about provision and marketing, given the international and direct-to-consumer NIPT markets.

*Although it would be feasible to restrict the advertising or sale of testing kits in the UK, it is not possible to prevent people from using services in other countries, such as those accessed via the internet.*

Clinical Genetics and Cytogenetics, Guy’s Hospital said that providers might be misrepresenting their products by describing them as ‘higher resolution’ tests which in reality do not perform better than other kinds NIPT.

*No. There appears to be very little regulation and given this is an extremely competitive commercial area, information provided to women and to clinicians can be misleading. Women are not always receiving the best test or correct information. Companies charge more for higher resolution tests that are not*

*fully validated or clinically indicated, whilst women may think that they are getting a better more comprehensive test.*

The DSA pointed to four-country variation in UK regulation of NIPT and called for a review of private screening. They cited the House of Commons Science and Technology [National Health Screening Report](#) conclusions that the General Medical Council and the Nursing and Midwifery Council should review their systems for ensuring good practice in this area.

*There are a number of ways in which private health screening is regulated in the UK and these vary from country to country. The DSA wants to see this reviewed. Mr Ben Gummer MP (former Parliamentary Under Secretary of State for Care Quality) stated in a letter to the DSA (27th April) that regulation in this area is not a simple landscape. The House of Commons Science and Technology Committee Report on National Health Screening also recognised this and recommended that “The bodies regulating the conduct of health professionals, including the GMC and Nursing and Midwifery Council review the effectiveness of their processes for ensuring that those operating in the private sector are providing patients with good quality, balanced information”.*

#### **4. NIPT provision**

Some concerns raised about regulation related to test provision and performance in the private sector. One area related to support made available for women and couples when they seek NIPT in the private sector, and the CMF argued that tests should not be provided without the involvement of a genetic counsellor.

*Private companies should not be able to sell NIPT direct to the public without ensuring the availability of trained genetic counselling services to recipients. The lack of such supporting services may diminish informed decision-making and render families vulnerable to targeting by unethical or fraudulent ‘providers’ and financial exploitation.*

The BMA took a similar view, advocating that a health professional should be involved with any form of genetic testing including NIPT, but was less optimistic about the feasibility of implementing such a policy.

*The BMA has taken the view that genetic testing via a health professional should continue to be the norm, and is preferable as accessing testing directly, without the input of a health professional, may not allow a discussion to take place as it would if part of an informed consent process. However we recognise that this cannot be enforced.*

The DSA stated that all healthcare professionals involved in screening, including NIPT, should be required to undergo training.

*The DSA is calling for the implementation of mandatory training for all health practitioners involved in the screening process for pregnant women in both*

*the public and private sector. Such training should be part of their professional qualifications.*

They cited their own [Tell It Right](#) training.

*The Down's Syndrome Association already provides a training module which could be used and further developed - TIR. The training is accredited by the Royal College of Midwives. Its key objective is to ensure health professionals have up to date, accurate information about living with Down's syndrome.*

## **5. Regulation of NIPT tests**

Genetic Alliance UK described the process via which NIPT tests are currently regulated, through the EU In Vitro Diagnostic Medical Devices Directive, pointing out that the requirements of the Directive focus on safety, rather than efficacy.

*NIPT, along with other types of genetic testing, is currently regulated in the UK under the [In Vitro Diagnostic Medical Devices Directive 98/79/EC](#). This requires all providers of genetic testing to register with the MHRA. However, the essential requirements aim to ensure that the products do not compromise the health and safety of patients and users, and there is no requirement to demonstrate the efficacy of a test.*

Future of Down's also suggested that current regulation was flawed and described the recent review of the Directive.

*It is widely acknowledged that the existing IVD directive 1998 is not fit for purpose. For this reason, the EU is currently overhauling existing medical devices legislation to strengthen the current approval system providing for stricter requirements for evidence of clinical performance and introducing stringent requirements for ongoing surveillance.*

Genetic Alliance UK suggested though that the replacement regulatory tool, the In Vitro Diagnostic Device Regulation, due to come into force in EU member states from May 2018, would impose burdensome requirements that may make it difficult for genetic prenatal tests to be provided through the NHS, but also observed that the UK's imminent exit from the European Union raised questions about the application of the regulation in the UK.

*The current regulations are soon to be replaced by the new In Vitro Diagnostic Regulation which was agreed in May 2016, and would be expected to become enforceable two years later. Amendments proposed as part of the drafting period include specific and onerous requirements on how genetic tests may be used, including providing that genetic tests may only be conducted by medically qualified personnel; that genetic counselling must be provided before and after a genetic test is delivered; and that an explicit written consent must be obtained before every genetic test is performed. This would prevent genetic tests being offered by genetic counsellors or midwives as part of*

*routine NHS prenatal care. Of course, the result of the recent referendum means that it is not clear to what extent the UK will be bound by the EU regulatory framework, or how much of it will be translated into UK regulations.*

Future of Down's added that the Medicines and Healthcare products Regulatory Agency (MHRA), the body responsible for regulating medical devices in the UK, do not consider the regulation of in vitro devices for which sample analysis takes place outside of the UK to be within their regulatory remit.

*NIPT regulation is further complicated by the UK MHRA declining to regulate IVDs where the analysis of the blood sample is conducted outside of the UK. This is based on the MHRA interpretation of the legislation wherein they consider the legislation to apply to the manufacturer not the end user. This interpretation neglects to recognise the companies...placing NIPT on the market within the UK (i.e., making the test available for paying prospective parents within a private clinic).*

This is problematic, they argued, since many NIPT samples are analysed outside of the UK, in regions where regulation may be minimal or not enforced, and women and couples undergoing NIPT in the UK may not be aware of this.

*For many NIPT systems the blood sample is taken from a UK patient and sent to a laboratory outside of the UK. In most circumstances this is the US. However, there is potential for a test laboratory to be located anywhere outside of the EU where regulation could be sparse to non-existent. The test and handling of an individual's genetic data are subject to local regulations. Whilst UK test centres offering NIPT using systems regulated outside the EU might state in small text at the bottom of a webpage that the system is regulated in the US under the CLIA exemptions, they do not in their advertising material make this clear the implications of this to prospective parents.*

Future of Down's also said that many NIPT samples collected from women in the UK are analysed in the US, where the Food and Drug Administration (FDA) controls medical devices, and pointed to issues with effective regulation in the US. It was noted that concerns about NIPT in the US had been expressed in the report, [The Public Health Evidence for FDA Oversight of Laboratory Developed Tests: 29 case studies 2015.](#)

*NIPT was amongst the 20 products identified... Their concerns include information citing high accuracy that is unsubstantiated, and the evidence that abortions had been undertaken based on results of false positives. They also reference the American College of Obstetricians and Gynaecologists statement in December 2012 that NIPT should not be offered to women in the general, low-risk population with resulting low PPVs.*

And made explicit the potential issues to which they thought inadequate regulation in the UK could give rise.

*Future of Down's has grave concerns over the MHRA decision not to regulate NIPT systems where the blood sample is tested outside of the EU. This position is vulnerable to abuse from manufacturers of NIPT who may in future seek to exploit this loophole and locate testing centres in regions where overheads are reduced and where there is the most lenient regulatory system... The potential for breaches of data protection are significant.*

Other respondents raised issues relating to the analysis of samples outside of the UK. The BMA pointed out that given that blood samples can be sent to laboratories anywhere in the world this created difficulties in controlling access to NIPT.

*As NIPT only requires a blood sample, in theory this can be taken, shipped and analysed overseas. NIPT may therefore be offered to consumers from anywhere in the world, making restrictions on what tests can be offered and regulations governing their use difficult, even if they were considered to be desirable.*

Genetic Alliance UK made the same point and drew parallels with cases in which women make use of PGD outside of the UK in order to avoid constraints on sex selection imposed by the Human Fertilisation and Embryology Authority (HFEA).

*Of course, due to the ease of shipping blood samples across borders, it is very easy for UK residents to bypass our stricter regulations and have their sample tested in another country. We are already seeing cases of women travelling to the US to have PGD sex selection for social reasons, for example, which is not permitted in HFEA licensed clinics in the UK, and a far more expensive and logistically challenging process.*

## **6. Legal issues concerning abortion**

The Church of England, Mission and Public Affairs Council raised more general legal issues relating to the Abortion Act and the use of information accessed using NIPT to help identify fetuses with Down's syndrome. They argued that section 1(1) (d) of the Abortion Act was not satisfied by pregnancies affected by Down's syndrome given that the condition is compatible with a high quality of life.

*...The current law permits termination of pregnancy if 'there is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped (Abortion Act 1967 [1d])'. Given the richness of life lived by very many people with Down Syndrome, it is highly questionable if this condition ought to be considered as a 'serious handicap', even though some people with Down Syndrome might have debilitating co-morbidities, as, of course, do others in society.*

Colette Lloyd said that given the need to undergo invasive prenatal diagnostic testing to confirm the results of NIPT it should be illegal for women to have an abortion on the basis of results of an NIPT test alone.

*Further termination based on the result of NIPT, and not on a diagnostic test, should be illegal.*

Genetic Alliance UK pointed out differences within the UK relating to abortion law and observed that it would be unlikely that the recommendation that NIPT be made available to women through the NHS would be adopted in Northern Ireland.

*Another regulatory issue relevant to NIPT is the laws in Northern Ireland around termination of pregnancy. At present in NI, abortion is not permitted even in cases where the fetus has a genetic abnormality incompatible with life. While the UK NSC makes recommendations which are applicable across the UK, it is up to each nation to decide when and how to put these into practice. Currently NI does not have a Down's syndrome screening programme but a second trimester fetal anomaly scan is offered. It would also appear unlikely that NI would implement NSC recommendations on NIPT.*

**We would like to identify the ethical values that are relevant or important in the context of NIPT. These might include: enabling informed decision making about reproduction, reducing harm, protecting the interests of future children, fair use of public resources, and the promotion of equality among members of society.**

**Question 19 - What ethical values do you think are important or relevant in the context of NIPT?**

### **1. Autonomy and informed consent**

Many respondents mentioned the value of informed consent, patient autonomy and women's reproductive choice in the context of NIPT. Clinical Genetics and Cytogenetics, Guy's Hospital said that "*informed choice and supporting women in their choices*" were important and Genetic Alliance UK said that "*the key ethical values here are informed choice and respect for the autonomy of the person.*"

ARC agreed that consent was a key principle, adding that this sometimes applied to a woman's partner too.

*As with anything pertaining to pregnancy, the principles of a woman's autonomy, informed choice and valid consent must be paramount – with inclusion of her partner in decision-making and care when appropriate.*

And a member of the BMFMS expressed this in terms of women's rights.

*Autonomy – the right for women to choose for themselves (informed consent).*

The Association of Genetic Nurses and Counsellors cited consent as important and emphasised the role for healthcare professionals in ensuring that this could be realised in practice.

*Informed consent – ensuring parents have been counselled by a suitably qualified healthcare professional about the process of NIPT and that all the possible findings (including the concept of incidental findings) are covered.*

A number of respondents pointed out that NIPT might also raise challenges for the possibility of informed consent. BioCentre echoed the perspective that reproductive choice was part of the value of NIPT but only if NIPT did not become 'routine'. The CMF said that "...*consent must be free of coercion*" and the DSA said "*the freedom to make an informed choice without duress*", rather than reproductive choice, as such, was the key value relevant to the use of NIPT.

The Liminal Spaces Project went further, pointing out that, in certain kinds of case, the availability of NIPT might positively undermine the prospects for women's reproductive autonomy. They highlighted the fact that NIPT may constitute a new, more effective means via which women might be coerced by others.

*What kind of additional burden might this put on, say, older women or women whose family or community practice non-medical sex-selective abortions? What does “choice” mean in this context and how meaningful might many parents’ or women’s choices be if the freer availability of such “easy” testing is, in fact, simply another means to coerce?*

The Liminal Spaces Project observed the nature of NIPT, as a blood test, and the role of private business in its provision might also impact on the possibility of genuinely free and informed choice. They said:

*One of the highest ethical priorities is to consider how the non-invasive nature of the test affects how (and how freely) decisions to undergo testing are made...The availability of such testing privately and with ease – potentially self-administered – raises important ethical and sociological questions around the patient-consumer tension.*

## **2. Value of human life**

Some respondents mentioned values relating to respect for, or sanctity of, life. The Anscombe Bioethics Centre said “...respect for the lives of all, irrespective of age, ability and location” and the CMF argued that:

*The value of a human life is not to be measured in terms of conformity to a genetic norm or by economic ‘productivity’, any more than by age (before or after birth), colour, race, gender or creed.*

The Church of England, Mission and Public Affairs Council raised ‘affirmation of life’ in a similar context and stated their view that, whilst abortion in their view may be acceptable in certain circumstances, any increase in the number of terminations should be viewed with this in mind.

*Affirmation of life: while recognising that NIPT will provide women with greater information during their pregnancies, as stated above, we are concerned that an unintended consequence of this might be an increase in terminations of pregnancy. The Church of England combines strong opposition to abortion with a recognition that there can be strictly limited conditions under which it may be morally preferable to any available alternative; such alternatives, however, require careful exploration.*

## **3. Equality**

A number of respondents raised issues relating to equality and inclusion. The Association of Genetic Nurses and Counsellors suggested that NIPT may raise issues of equality for those with the conditions for which NIPT is used to test.

*Promotion of equality among members of society – prenatal testing is already offered for these conditions. However, as NIPT is ‘just a blood test’, it may increase the uptake of testing and then the number of terminations for these*

*conditions, which does not promote equality for individuals with these conditions.*

BioCentre also raised principles relevant to the wider societal implications of NIPT and emphasised the value of compassion in the context of equality, suggesting that NIPT may undermine aspirations to make society more equal.

*A compassionate society rejects reductionism and considers everyone as equally valuable based on who he or she is and not what he or she can do. Whatever good intentions may be advocated, permitting practices that inevitably make a distinction that some people are of lesser value than others and ought to be screened out before birth should be opposed.*

CARE made a very similar point.

*The most important value to have in mind when considering NIPT is that preserving the compassionate nature of our society depends upon viewing everyone as equally valuable and of not permitting practices that, despite good intentions, will inevitably have the effect of conveying that some people are of less value and can be screened out before birth.*

DSA cited inclusion as a key principle and argued that the social model of disability was also important

*A commitment to the social model of disability...a commitment to inclusion*

Another aspect of claims about the importance of equality was stressed by the CMF who suggested that the potential for NIPT to be used for sex selection may undermine aspirations for gender equality.

*The high degree of accuracy of NIPT as early as seven weeks of pregnancy carries a number of socio-ethical implications, such as the selective termination of fetuses according to sex, in communities where it is culturally desirable to have male offspring.<sup>13</sup>*

#### **4. Reduction of harm**

Avoidance and/or reduction of harm was mentioned by a number of respondents. Clinical Genetics and Cytogenetics, Guy's Hospital said that the principle of "...do no harm" was relevant and the Association of Genetic Nurses and Counsellors said:

*Reducing harm – by offering NIPT, this should reduce miscarriages caused by invasive testing. Some may argue that this testing may cause less psychosocial harm to patients, as it is a blood test and can be carried out earlier in pregnancy.*

---

<sup>13</sup> See CMF's full consultation response for references.

BPAS agreed that harm posed by miscarriage was of key importance and expressed the view that this would be seen as the determinant factor for most people.

*NIPT will provide pregnant women with more information to make choices in distressing situations. The risk of miscarriage is of grave concern to women needing further antenatal testing and the availability of a test which prevents some patients needing to take that risk is a positive step forward.*

Respondents had different ideas about what the implications of a harm reduction principle might be in the case of NIPT though. A member of the BMFMS agreed that "...first do no harm" was a key principle but said that its relevance to NIPT related to "the risks of causing anxiety and terminating wanted pregnancies based on fear and uncertainty." Future of Down's also perceived the principle of harm minimisation to be important but said that there were also risks of harm for women and couples.

*In opting to terminate parents often express concern for existing siblings, and concerns that pressure from the added stress of caring for a child with Down's syndrome will have a negative impact on their relationship. The irony is that in the case of Down's syndrome a medical intervention to deselect a previously wanted pregnancy carries a risk of harm to the parents that may be more likely to realise these concerns, whereas evidence from studies of wellbeing characteristics in families with offspring who have Down's syndrome support assertions from parents that parenting a child with Down's syndrome is on the whole a positive experience.*

Colette Lloyd thought that application of a harm reduction principle implied that NIPT should not be used to test for Down's syndrome since the potential for increased terminations posed the risk of harm to fetuses.

*Reducing harm: The relevant question is "reducing harm against who?" The RAPID screen, using outdated figures of risk for amnio/CVS (0.5%) predicted a reduction in miscarriage of 25. Up to date figures from Akolekar's (2015) systematic review (0.1%) would produce a reduction of 5 miscarriages. However, NIPT is also estimated to detect an increased 102 pregnancies with Down syndrome, and with the current 90% termination rate, that would be an increase of loss of life of approx 85. Reduction of harm would be achieved by: not introducing this NIPT into the NHS; introducing a ban on termination due to Down's Syndrome; and better education of health professionals so that all give a non-discriminatory view of trisomy 21.*

## **5. Interests of future people**

A range of ethical considerations cited by respondents related to the interests of people that fetuses may become. The Association of Genetic Nurses and Counsellors observed that information revealed by NIPT about late onset conditions in particular raised issues in this area.

*Protecting the interests of future children – there are a number of adult onset conditions (for example Huntington’s Disease) where we know that many adults do not opt for predictive testing. If babies were born who had tested positive for such conditions (through NIPT), this removes their autonomy to choose whether or not to know.*

BioCentre also raised ethical issues with future children being made aware of serious health conditions they would go on to develop, arguing that there “are no short-term benefits either to a child knowing she will (or is very likely to) develop an adult-onset genetic condition or to the parents knowing this.” The value of an open future was cited as a key principle in this context by BioCentre who described the potential harms of violating this principle.

*Advocating an ‘open future’. As a consequence of parents choosing to undertake the testing, it violates the child’s ‘open future’<sup>14</sup> to decide for themselves what tests to have. Rather than feeling psychologically prepared, the child may feel greater anxiety knowing she faces a future with a particular condition.<sup>15</sup>*

Colette Lloyd also said that “protecting the interests of future children” was a relevant principle but linked this point with one about abortion and philosophical issues concerning attempting to weigh the interests of ‘individuals’ who are not brought into existence. She said that people were wrong to think that they might be acting in the interests of a future child by using NIPT and selective termination to avoid giving birth to a baby with a genetic condition, given that that child would not exist.

*Protecting the interests of future children: Again, exactly whose interests are protected here? If pregnancies are terminated as a result of this testing, then any children who come to exist will be different children. The temptation is to think that children will be better off if they don’t have Down syndrome. This in itself is highly controversial, but even if it were true, if the particular future children being tested no longer exist, then their interests are not protected; they are stopped from having interests. A similar philosophical mistake is at work when we say things like, “It would have been better for me if I had lived in Victorian times,” forgetting that anyone who lived in Victorian times would not have been you, and therefore this could not actually be in your interests.*

Many of the issues connected to the interests of future people, the Liminal Spaces Project said, related to the right not to know about one’s genetic makeup. They said that this should be understood as an issue of privacy rather than autonomy, which would make European legislation on the right to respect of one’s private life applicable.

*...In circumstances where termination is not chosen (or is not an option), then what we are really concerned with are the rights and interests of the potential future child....We have argued elsewhere that there is a need for more*

---

<sup>14</sup> See BioCentre’s full consultation response for references.

<sup>15</sup> See BioCentre’s full consultation response for references.

*conceptual clarity about the nature of claims around the putative ‘right not to know’, in particular that it is conceptually more coherent to see this as an aspect of privacy protection rather than autonomy protection.<sup>16</sup> Put simply, while autonomy is about self-determination (and so implies that one is capable of acting autonomously and that one can positively choose between options), privacy is about a state of non-interference (by the state or others through horizontal effect), and this includes the imposition of unwarranted or unsolicited information. Consider the example of various international instruments such as the Oviedo Convention that purport to recognise the right not to know. They do so, however, by saying that a person’s wishes not to know should be respected. This assumes, however, that (a) a person is capable of exercising that choice [not so with fetuses or young children], and (b) that the person knows that there is something to know. But what if you do not know that there is something to know, or you cannot exercise such a right? How does autonomy help? It does not. Rather, we would argue that privacy - as embodied in human rights globally, nationally and in the European Convention on Human Rights (ECHR) - recognises a fundamental (legal) right to respect for your private life, and this means a prima facie presumption against interference, including – we suggest - imposition of unsolicited information unless there are good reasons to provide it (or generate it at all).*

These issues also related, they said, in part to potential “*discrimination or stigmatisation for future children being ‘labelled’ in some form or another because of a choice made before their birth and without due and full regard to the consequences for their lives.*”

## **6. Fair use of resources and equitable access**

Some appealed to the principle of fairness in resource allocation in connection with the proposal that NIPT be made available on the NHS. Clinical Genetics and Cytogenetics, Guy’s Hospital cited “*fair use of public resources*” and the Association of Genetic Nurses and Counsellors said:

*Fair use of public resources – this is always an issue due to the limited funding for the NHS. We do not know how costs compare with current screening so cannot comment on this.*

Equitable access was also seen as an important, relevant value and Clinical Genetics and Cytogenetics, Guy’s Hospital said “*equity of access*”.

## **7. Objective of medicine and conscientious objection**

The Anscombe Bioethics Centre raised ethical issues relevant to those working in healthcare professions, specifically. They said that the goal of medicine to promote health was a relevant value. They also said that the entitlement of healthcare professionals to conscientiously object to healthcare practices with which they do not agree was a relevant principle.

---

<sup>16</sup> See Liminal Spaces Project’s full consultation response for references.

*Respect for the healthcare professions as properly centred on promoting health and not on social exclusion or quality control of existing human lives...the protection of healthcare professionals from pressure to participate in NIPT.*

## **8. Care of the vulnerable**

Care of the vulnerable was seen by the Church of England, Mission and Public Affairs Council to be an important relevant principle

*Care of the vulnerable: we stress that women who are informed that NIPT suggests that there is a high probability of a chromosomal abnormality being present are vulnerable and require further information, support and counselling as they undergo diagnostic tests.*

## **9. Legal issues**

Saving Down Syndrome pointed to a number of areas of law that they said were relevant to ethical values pertaining to NIPT.

*It is not for us to comment on the ethics of individuals but we do consider it appropriate to call for the recently developed Human Rights narrative around disability to be invoked for NIPT. Therefore the ethical values of the UK and the UN are obviously the values which will be relevant and important to NIPT, and have to be addressed without further ado:*

- *The Equality Act 2010*
- *The United Nations Convention on the Rights of People with Disabilities*
- *The United Nations Bioethics Committee Report, Updating its Reflection on the Human Genome*
- *The landmark Montgomery Case*

CARE also mentioned the United Nations Convention on the Rights of People with Disabilities (UNCRPD) and added that the UN Convention on the Rights of the Child (UNCRC) was relevant.

*The UN Convention on the Rights of the Child (UNCRC) states that a child “needs special safeguards and care, including appropriate legal protection, before as well as after birth”. During an Office of the United Nations High Commissioner for Human Rights debate on how the UNCRC applies to disabled children, the following was noted: “All children were equal members of the human race, discriminatory laws which denied their right to life should be repealed...It was one thing to work to eliminate impairment but quite another to eliminate the person with the impairment... Rather, we must celebrate diversity and learn to celebrate the birth of every child, with or without disability.”...The UN Convention on the Rights of Persons with Disabilities (CRPD) has a principle, the “respect for difference and acceptance of persons with disabilities as part of human diversity and humanity” ([UN Convention on the Rights of Persons with Disabilities](#) (2006) Article 3). In*

*Concluding Comments on reports from Spain and Hungary, the Committee on the Convention called for action to prevent discrimination within abortion law on the grounds of disability.*

The Montgomery case was mentioned by a number of respondents. The CMF said:

*Consent must be fully informed and all the more so following Montgomery v Lanarkshire Health Board. This requires pre-test and post-test information and counsel by trained staff, able to give neutral advice. Health professionals should signpost families receiving a diagnosis of disability to information leaflets covering all their options, to telephone and online helplines manned by trained professional counsellors, and to local and national support groups for those with specific conditions. Following her decision, and regardless of what choice the woman and her family may make, ongoing support must be part of that provision.*

The Public Sector Equality Duty was raised by Lorna Watson.

*The testing as it stands contravenes the justice principle as applied to public health programmes (Schroder-Back). It needs to be examined in the context of the Public Sector Equality Duty. It is misunderstood to be a public health objective to reduce birth prevalence of Down's syndrome based on cost savings but this is not a legitimate aim for public health, as people with Down's syndrome have equal rights to be valued members of society.*

## **10. Ethical status**

Some respondents used this section to assert views about the overall ethical status of use of NIPT. BPAS said:

*There will be a minority of voices who do not believe that it is ethical to offer NIPT. The majority will consider the offering of NIPT to pregnant women with a risk of fetal anomaly is profoundly ethical and will trust women to make their own decisions in difficult circumstances.*

ARC said that no new issues were raised by NIPT.

*I do not feel that NIPT as a technology does more than heighten our awareness of current ethical principles which should apply to any prenatal testing.*

## Question 20 - Any other comments: Please tell us anything else you would like to raise in relation to NIPT

### 1. Issues relating to informed consent

Some respondents here emphasised the importance of ensuring women are able to make informed decisions about whether to undergo NIPT, and the need for adequate NHS resources to ensure this is possible was stressed by one anonymous respondent. Suggestions for ways of improving consent processes were also made and Colette Lloyd proposed that use of visuals, videos and graphics including a “flowchart pathway for where the decisions to test may lead to further, more life changing decisions” would be useful. She also described a possible consent protocol designed to ensure that undergoing prenatal testing is the result of an informed choice.

*A potential method of ensuring informed choice about testing can be found in the case of the newborn screen for Duchennes Muscular Dystrophy. Parents were given the opportunity to post off the blood draw themselves if when they arrived home they decided that they did really want to have the test. A similar practice in the case of NIPT would ensure women were making a conscious decision about whether they wanted testing (Parsons et al, 2000).*

The DSA also stressed the importance of consent, highlighting the need for quality information and training, the development of which they said would benefit from their own involvement.

*The Down’s Syndrome Association can make a valuable contribution in both the development of information for women and training for health care professionals. It would like to work with the UKNSC and the NHS, in the design, development and delivery of any modules of training being considered by the UKNSC.*

### 2. Need for support and advice

Some respondents used their comments to emphasise the different areas where women and couples should be provided with support. The CMF called for adequate support for disabled people and their families.

*Bringing up a child with special needs often involves substantial emotional and financial cost for families. Practical support for the longer term must be in place for families, and access routes to financial, emotional and practical support as well as treatment need to be clearly signposted. These should include routes for exploring adoption for those families who feel personally ill-equipped but who wish to offer their child ‘the gift of life’. More statutory funding should be provided for information, care and support groups and organisations for those with disabilities.*

ARC made comments on the risks associated with the private provision of NIPT and suggested that the Council should make recommendations on how women and couples can be advised and supported to identify quality service providers, and ensure that effective scrutiny and oversight is in place for clinics and hospitals providing NIPT.

*What is perhaps worth noting about NIPT (also true of assisted reproduction technologies) is the fact that private provision is always going to outstrip what is available on the NHS. In view of this, ARC would like to see the Nuffield Working Group make strong recommendations as to how we can assist expectant parents, wishing to access private services, to seek out responsible providers and how best to expose those looking to cynically exploit a lucrative market.*

### **3. Implications for prenatal screening**

Some thoughts were expressed on the possible implications for other areas of prenatal screening of making NIPT available on the NHS. A member of the BMFMS suggested that serious consideration would need to be given to the issues raised by the possibility of de-skilling in the field of invasive diagnostic procedures. They suggested that a 'roving' service might be needed in order to address the potential shortfalls that may arise in skills of invasive testing.

*While the private implementation may not reduce invasive testing much, widespread NHS availability will reduce the need for invasive testing dramatically. At this point, now, we need to start having dialogues about provision of invasive...or a roving service. Where regions involve more than one health board and deanery this discussion will take some time. There are implications for training and other ultrasound guided needle procedures: clinicians providing more advanced procedures such as fetocide, cordocentesis and laser ablation have traditionally relied on a large experience of amnio then CVS beforehand and no longer will have these skills. There are implications for workforce planning and subspec training, perhaps we may have a situation where not all subspec trainees do needle procedures. There will need to be established role of validated simulator training.*

On the other hand, another member of the BMFMS said that the introduction of NIPT within NHS antenatal care could give rise to increased pressures in other areas of NHS prenatal screening.

*In a few years women will have voted with their feet. Those that can afford it will have NIPT straight off leaving those that can't to have NHS. The rise in women going for screening of up to 150% (65% uptake to near 100% (as being forewarned without risk will become a viable option) will put further demands on overstretched scan departments where sonographers are in short supply and those we can keep in post, develop RSI from repeated NT measurement.*

#### **4. Conflicts of interests**

Colette Lloyd raised concerns about the conflicts of interest raised by the fact that significant amounts of money had already been invested by different parties in NIPT technologies. She said that some private companies had made considerable investments in technology used in analysing DNA.

*Return on financial investment must not be allowed to be a driving force in this debate.*

She added that endeavours to show that widening access to NIPT has been cost effective should not manifest as pressure on women to make use of NIPT.

*In the past there has been a desire amongst people providing screening for any condition to increase the uptake of the test (Raffle, 2001), which will in turn increase cost-effectiveness. It is extremely important that this attitude does not continue to prevail in the case of prenatal screening tests. Women must be clearly informed of the limitations of the test, and not pressurised in any way to have it.*

#### **5. Ethical issues**

Ethical issues were raised explicitly by Colette Lloyd who said that “...there seems to be a rush to bring this test into the NHS, the go-ahead for an evaluative roll-out before ethics had been fully considered is an example of this.” An anonymous respondent said though that recent legal decisions suggested that there was a duty on healthcare professionals to ensure that women were informed about the existence of NIPT, adding that not giving women access to NIPT through the NHS was ethically problematic.

*Legally the Montgomery vs Lanarkshire ruling in the Supreme Court means all pregnant women are entitled to receive the material facts about NIPT. Limiting access to NIPT to those who can afford it would be a further example of inequity in health provision.*