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ARTICLE

Non-invasive prenatal diagnosis for fetal sex determination: benefits and disadvantages from the service users' perspective

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Prenatal fetal sex determination is clinically indicated for women who are at risk of having a child with a serious genetic disorder affecting a particular sex. Ultrasound has been the traditional method used, but early fetal sex determination using non-invasive prenatal diagnosis (NIPD) can now be performed using cell-free fetal DNA in maternal plasma. The study aim was to assess the views and experiences of service users who had used NIPD for fetal sex determination. In this paper, we report on the perceived benefits and disadvantages. A qualitative approach using semi-structured interviews was used. A total of 44 participants (38 women and 6 partners of participating women) were recruited. Participants' views and experiences of NIPD were overwhelmingly positive. Concerning benefits over traditional methods, three themes emerged: (1) technical aspects of technology; (2) timing; and (3) enhanced decision-making. Practical advantages of NIPD included avoiding miscarriage, and there were a number of psychological advantages associated with timing such as perceived control, early re-engagement, normalization of pregnancy and peace of mind. Participants also valued NIPD as it enabled a stepwise approach to decision-making. A number of disadvantages were discussed including concerns about social sexing and increased bonding at a time in pregnancy when miscarriage risk is high. However, participants felt these were fairly minor in comparison with the advantages of NIPD. Until definitive genetic diagnosis using NIPD is available, NIPD for fetal sex determination is perceived as a good interim measure with a number of notable advantages over traditional methods.
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INTRODUCTION

Prenatal fetal sex determination is clinically indicated for women who are at risk of having a child with a serious genetic disorder affecting a particular sex. This includes women who are carriers of X-linked genetic disorders such as adrenoleukodystrohy (ALD) and Duchenne muscular dystrophy (DMD), where fetal sexing is often used to guide decisions about invasive testing, or carriers of hemophilia, where it can inform management of labor and delivery of 'at risk' male pregnancies. In addition, fetal sex determination is used for conditions associated with ambiguous development of the external genitalia, such as congenital adrenal hyperplasia (CAH), where maternal steroid treatment early in pregnancy can reduce the level of virilisation in female fetuses.[1]

Ultrasound has been the traditional method used for fetal sex determination. In the second and third trimesters, it is accurate in >99% of cases with normal genitalia.[2] Early ultrasound (12–14 weeks) is also a reliable option when performed at specialized centers.[3–5] Invasive testing, either using chorionic villus sampling from 11 weeks or amniocentesis from 15 weeks,[6–8] is also an option and allows definitive genetic diagnosis, but both techniques carry a small but significant miscarriage risk (~1%).[9]

When used for determination, NIPD has a number of advantages over ultrasound and invasive testing. It is feasible from 7 weeks' gestation,[1,10,11] has been shown to be >99% accurate[11] and as it is non-invasive, carries no risk of miscarriage. Furthermore, it reduces the need for invasive procedures by up to 50%[1] and has also been shown to be cost neutral for those conditions, such as DMD, where the majority of carriers choose to have invasive testing.[12]

Although the clinical value of NIPD for sex determination is clear, little is currently known about service users' views and experiences of the test. In this paper, we report on the benefits and disadvantages of NIPD from the service user's perspective. Service users' experiences of and preferences for service delivery are described elsewhere.[13]

METHODS

Approval for this qualitative study was obtained from the NHS Research Ethics Committee (10/H0724/41) in June 2010. Participants were recruited through one hemophilia clinic, two fetal medicine units and two genetic centers that were located in either London or the South of England. A member of staff identified women and partners who had used NIPD for fetal sexing and they were sent an invitation letter from the clinic and a patient information sheet describing the study.

Face-to-face or telephone semi-structured interviews were conducted by the lead researcher (CL). Interviews were chosen as the primary method of data collection as they enable in-depth exploration of the subject matter and can uncover new areas not anticipated at the outset.[14] The interview schedule was semi-structured and explored issues including: their reasons for using NIPD; the perceived value and disadvantages of NIPD; their experience of taking the test and receiving the test results. Where partners had agreed to be interviewed,

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in all cases participants had a preference to be interviewed together. Interviews were audio recorded, transcribed verbatim and anonymised.

For the more structured questions, data analysis was conducted using thematic analysis.[15] For the exploratory questions, the methods described by Strauss and Corbin[16] were used in order to ensure themes emerged inductively from the data. The software package NVivo version 8 (QSR International, Pty Ltd, 2008) was used to facilitate data analysis. Data collection and analysis was done concurrently. Transcripts were read repeatedly by CL and broken down into small meaningful units of text. As coding continued, codes were clustered to form broader categories. To ensure inter-rater reliability, a subset of transcripts were read and coded by two other researchers (MH, CC). Any discrepancies were discussed until consensus was reached. During the coding process, emerging themes were worked back into the topic guide to explore further during interviews. At the point at which no new themes were emerging from the data (saturation), recruitment ceased. The themes were then examined to identify relationships between them.

RESULTS

Participant characteristics

A total of 87 women were approached to participate in the study. In all, 45 participants (38 women and 7 male partners of participating women) representing carriers and in 3 cases potential carriers (as carrier status was not confirmed), agreed to take part in the study (44% recruitment rate). A total of 10 genetic conditions were represented (Table 1). Overall, 27 interviews were conducted face-to-face and 11 by telephone. The interviews lasted for 20–55 min. Participants were interviewed between October 2010 and August 2011. All 38 women had undergone NIPD for fetal sex determination. Some had used it in more than one pregnancy, giving a total of 61 pregnancies for respondents. In 19 pregnancies, women had gone on to have invasive testing for definitive diagnosis (Table 2).

Table 1 Demographic characteristics of participants

Disease	N = *38*
Hemophilia	19
Duchenne muscular dystrophy	5
Adrenoleukodystrohy	4
Congenital adrenal hyperplasia	3
Androgen insensitivity syndrome	1
Becker muscular dystrophy	1
Charcot-Marie Tooth disease	1
Pelizaeus-Merzbacher disease	1
X-linked severe combined immunodeficiency	1
X-linked mental retardation	1
X-linked hydrocephalus	1
Age	N = *44*
18–21	1
22–25	1
26–29	9
30–33	13
34–37	10
38 +	10
Ethnicity	N = *44*
White	42
Black or Black British	0
Asian or Asian British	2
Mixed	0
Other	0
Education	N = *44*
Secondary school – O level/GCSE	12
Secondary school/college – A level	12
University first degree	13
University higher degree	7
Has an affected child	N = *38*
Yes	18
No	19
Don't know	1

Interview findings

Participants' views and experiences of NIPD for fetal sex determination were overwhelmingly positive with words including 'brilliant', 'exciting' and 'incredibly lucky' being used to summarize the experience. Those participants that received news that the fetus could be affected, including those that went on to have IPD, were found to be equally positive towards the technology, as those who were told that the fetus was of the unaffected sex. The fact that a number of participants used NIPD on more than one occasion indicates the value that the participants placed on early information with no risk to the fetus. Concerning the benefits of NIPD, three specific themes emerged, which comprised both practical and psychological benefits. These were: technical aspects of testing procedure; timing; and enhanced decision-making. Some disadvantages of NIPD were also raised including: increased bonding between mother and fetus when miscarriage risk is high; increased anxiety; connection to a fetus when the pregnancy may be terminated; robbed of surprise; and misuse of technology.

Technical aspects of testing procedure

Safety. Safety was found to be a key value of NIPD. For those women who had experienced both non-invasive and invasive testing, the advantage of NIPD was that the procedure posed no risk to the mother or fetus.

> 'Having an amniocentesis, obviously there's a risk of miscarriage. With the non-invasive blood test there's none of those associated risks so it's much better.' Di – CMT carrier

Decision-making around invasive testing was identified as a distressing choice participants had to make owing to the associated risk of miscarriage.

> 'Being told that you are doing something that could risk the pregnancy, it's your choice and you are choosing to have a test, that's quite distressing.' Zoe – X-SCID carrier

In comparison, no such emotion was experienced by participants when deciding whether to use NIPD.

Ease of the testing procedure. NIPD was acknowledged to be an 'easy', 'quick' and 'simple' test to conduct as it was a blood test. In comparison, invasive testing was described as 'surgical', 'stressful' and 'painful'. Some women discussed how NIPD was psychologically 'easier' than invasive testing as there was no visual acknowledgment of the fetus during the testing procedure, unlike with invasive testing.

> 'I think that is quite a difficult situation actually being able to see what's going on while the actual test is being done.' Sarah – hemophilia carrier

Seeing the image of the fetus on the ultrasound monitor was found to be distressing; in one case this had caused a participant to change her mind about proceeding with invasive testing after NIPD.

Table 2 Information around NIPD experience

Years since first used NIPD	N = *38*
<1	2
1–2	9
2–3	10
3–4	2
4–5	6
5–6	5
>6	2
Don't know	2
Number of times used NIPD	N = *61*
1	24
2	9
3	5
4	1
Results from NIPD	N = *61*
Fetus of unaffected sex	32
Fetus could be affected	29
Pregnancy outcomes	N = *61*
No further testing	37
Invasive testing (termination)	19 (7)
Miscarriage	5

Abbreviation: NIPD, non-invasive prenatal diagnosis.

Women commented that during pregnancy numerous blood tests were conducted, thus this test was no different. There was, however, an acknowledgment that despite the ease of the procedure, the results could have important consequences.

> 'Having that blood test is just nothing, it's like any of your other visits to hospital when you're pregnant, it's just the results that have such a big impact.' Lara – hemophilia carrier

Timing

Knowing the sex of the fetus from around 9 weeks gestation was identified as being advantageous by all participants. Nevertheless, a number of specific themes surfaced during the analysis.

Preparation. The results of NIPD enabled participants to prepare both practically and psychologically for the next stage of the pregnancy. Women going on to have invasive testing could ensure arrangements were put in place at the earliest possible opportunity. There was also more time to mentally prepare oneself for the possibility of having to terminate the pregnancy.

> 'I think it gives you time to prepare … I feel that I had some time to go through the stage of getting upset and crying, getting emotional before the amnio and then going, no this is what I'm doing. So having those few weeks was really valuable.' Fay – DMD carrier

Decision-making around invasive testing and termination was identified as being easier in the first trimester, before the pregnancy was physically obvious to others and fetal movements were felt.

> 'I knew it wasn't really much of a baby inside me. So it was easier to make a decision about termination.' Gail – X-linked mental retardation carrier

For those participants who had chosen to continue with their pregnancy without going on to invasive testing, the information could be used to inform where the baby was delivered (hemophilia), the need for continued steroid treatment (CAH) or allow time to prepare for possibly having a (or another) child affected by the condition (hemophilia, BMD).

Control. Control emerged as an important theme within the data with NIPD empowering women to regain, at an early stage, a sense of control over their pregnancy.

> 'It's almost like you feel you've got a bit more control, you haven't got control, but you're not waiting for months … You do feel good that actually you can dot the I's.' Trish – DMD carrier

Although the gender information gained from NIPD was not necessarily what participants had been hoping for, the increased sense of control outweighed any additional anxiety the information generated.

> 'I do think you have to arm yourself with as much knowledge as possible. Yes it's unbearable, but at least you know as much as you can possibly know at that time.' Ruth – ALD carrier

By virtue of being a carrier, there was an expectation from many women at the outset that the pregnancy would not necessarily run smoothly; hence, control through information may have been something they actively pursued.

> 'I was happy to know by nine weeks just to know where I was going, even if it was bad news, because at the end of the day I knew I was a carrier so it wasn't unexpected.' Jade – Pelizaeus Merzbacker disease carrier

Peace of mind. Knowing the sex of the baby early on was found to impart a feeling frequently referred to as 'peace of mind'. Where the fetus was identified as female for X-linked conditions, no further testing was necessary enabling parents to 'just relax and get on with enjoying the fact you're pregnant'. Rose – ALD carrier

This sense of relief early in pregnancy was accentuated for those participants who had experienced invasive testing and a termination of pregnancy in a previous pregnancy.

> 'It actually did make a big difference because it was after a termination and it was like, we've got to go through all this again and actually we didn't have to go through it all, we only had get to nine weeks, have a quick blood test and then everything is over really and we could continue as normal … we could heave a sigh of relief.' Sarah – hemophilia carrier

When the fetus was identified as 'at risk', the information was still found to provide peace of mind to those participants continuing with their pregnancy. For hemophilia carriers, there was reassurance in knowing that the pregnancy was being monitored and that delivery would take place at a specialist center. For carriers of CAH, if the fetus was female there was peace of mind knowing that they were taking appropriate medication. If the fetus was male, they could stop taking the steroid medication altogether.

> 'For me, I could stop my treatment, I could relax about my baby not having the physical differences, not needing the surgery or anything, so it was very much peace of mind.' Anna – CAH carrier

Delayed attachment to pregnancy. It became apparent that there were mixed emotions regarding pregnancy among the participants, for whom termination was an option, with a number of women who had purposely tried to remain detached from their pregnancies until the risk of having an affected child was no longer present, as a form of self-protection.

> 'You don't want to get attached because you don't want to make it any harder on yourself.' Jodie – X-linked hydrocephalus carrier

For those participants who received news that the fetus could be affected, the anxiety and disengagement was prolonged. Yet for those that received news that the fetus was of the unaffected sex, the anxiety dissipated and participants described feeling 'relieved', 'happy' and 'excited'. Furthermore, they were able to re-engage with their pregnancy at an earlier stage than if they were relying on the sexing scan or invasive testing.

Normalization of pregnancy. Women were aware that during the first trimester, there was an increased risk of miscarriage and for that reason, their own risk and anxiety was compensated for to some extent because other pregnancies were also at risk during that period.

> 'We sort of said, 'we're in the same boat as everyone else because there are all sorts of things that can go wrong, especially in the first few months.' Lara – hemophilia carrier

When the fetus was identified as female for carriers of X-linked disorders (or male in the case of CAH) through NIPD, there was no longer an additional risk above and beyond what most people experienced during the first trimester. NIPD therefore allowed women to feel they were having a 'normal pregnancy' at an early stage and were able to tell family and friends at a time of their own choosing.

> 'The blood test I suppose was quite nice in the fact that after that I only had to get to the twelve weeks like any other pregnant woman and that was it.' Jade – Pelizaeus Merzbacker disease carrier

In comparison, those at increased risk opting for invasive testing had increased anxiety and the need for secrecy around the pregnancy was prolonged.

Decision-making

A further value of NIPD was as a facilitator of decision-making. There were a number of factors that were found to have impacts on this, including: whether a treatment was available; pressure from other family members; which pregnancy it was; previous experiences during pregnancy such as miscarriage or termination; personal experience and perceived seriousness of the condition.

> 'My brother all his life has been basically a human pin cushion … Right throughout my childhood I've always seen him with wires, on a life support machine, you know, so I've seen all that.' Melissa – ALD carrier

As a result, decision-making was identified as a dynamic and sometimes complex process. Participants broadly fell into one of the three categories (Table 3).

Continue without further testing. A number of participants had already decided (either before pregnancy or before receiving the NIPD results) that they would continue the pregnancy, whatever the sex of the fetus is. These women were carriers of conditions that were considered by participants to be treatable, and hence not severe enough to warrant termination of pregnancy. Here, the main reason for taking the test was to inform delivery or, for carriers of CAH, manage steroid treatment. The advantage of NIPD over ultrasound was that the information was highly accurate and was available earlier.

Invasive testing if necessary. Participants in this group had decided before NIPD that they would undergo invasive testing and termination if necessary. In many cases, these participants had had a child or other family member affected by the condition. For these participants the main benefit of NIPD was avoiding an unnecessary invasive test. Two carriers of CAH indicated that they would have taken an invasive test if the fetus was female to identify whether it was affected, but would not have gone on to have a termination of pregnancy. Anna spoke about the importance of knowing in order to 'set up a birth plan because the baby has to be given some treatment' (Anna – CAH carrier). For Sharon, the main reason for wanting to know was mental preparation:

> 'I am very much the school of thought of be prepared kind of thing, rather than it be sprung on me.' Sharon – CAH carrier

Table 3 Types of decision-making

Types of decision-making	*Condition*	*Example quotes*
Continue without further testing	Mild to moderate hemophilia, CAH.	'When I first discussed it with them, when they sort of said, 'oh a test to sex the baby' and things, I thought it was an invasive test and I knew I didn't want to do that … I wouldn't be willing to, sort of, terminate a pregnancy.' Linda – hemophilia carrier
Invasive testing if necessary	Moderate to severe hemophilia, DMD, ALD, x-linked hydrocephalus, X-SCID, Pelizaeus-Merzbacher disease, AIS, X-linked mental retardation.	'I knew I was going to have an invasive test if it was a boy.' Susan – hemophilia carrier. 'Dad finally got diagnosed with adrenoleukodystrophy and that was an exceedingly unpleasant period in his life. It was absolutely horrible … You wouldn't put your worst enemy through that … so I would have had the invasive as I wouldn't have a boy with ALD.' Ruth – ALD carrier
Undecided	Mild to moderate hemophilia, moderate to severe hemophilia, BMD, CMT.	'Each baby step was so helpful in having the discussions. I think with the second one, if I'm honest, I think I'd have been more likely to terminate. With the third one, whether it's because I'd had two miscarriages, you start to question the whole thing … but again, you can only reach that decision once you've taken those steps. I couldn't have said that at the start.' Cathy – hemophilia carrier 'And I think that's the key is that once you know the sex you can either put it to the back of your mind or think right now I've got to go through the next stage and I think you can take it more in bits and pieces rather than it all in one go.' Trish – DMD carrier

Abbreviations: AIS, androgen insensitivity syndrome; ALD, adrenoleukodystrohy; CAH, congenital adrenal hyperplasia; CMT, charcot-Marie Tooth disease; DMD, Duchenne muscular dystrophy; SCID, severe combined immunodeficiency; X-SCID, X-linked severe combined immunodeficiency.

Undecided. Some participants were undecided at the time of testing as to what choices they would make. This decisional delay occurred for a number of reasons including differences in opinion between partners, pressure from a family member with the condition, desire to delay decision-making until the gender of the fetus was known, and awareness that feelings towards invasive testing and termination were likely to change during the course of the pregnancy.

The option of NIPD was found to enhance the experience for this group as it enabled them to adopt a stepwise approach to decision-making. This approach allowed them to create distinct timeframes by dividing time into manageable 'chunks' and focus on the immediate future. This strategy is highlighted by Trish who describes making decisions in 'bits and pieces' (Table 3).

Disadvantages of NIPD

When asked about possible disadvantages of NIPD, five themes emerged: miscarriage risk; increased anxiety; connection to a fetus when the pregnancy may be terminated; being robbed of a surprise; and misuse of technology.

NIPD increases bonding at a time when miscarriage risk is high. Five participants experienced a spontaneous abortion after they had received their NIPD results. In two of these cases, participants felt that, as a result of knowing the sex of the fetus (which in both cases were female), an identity and connection with the fetus had been established.

> 'The only disadvantage I can think of from personal experience is because we lost the girl that we were carrying. Obviously I'd started to bond and we had a name and it seemed much more real.' Marie – hemophilia carrier

Two of the participants (who had both decided to undergo invasive testing before losing the pregnancy) maintained that when weighing up the risks and benefits, they still valued the test results despite the increased psychological engagement in the pregnancy as a result of knowing the sex, at the gestational period during the first trimester when miscarriage risk is elevated.

Increased anxiety. Women going on to have IPD experienced increased anxiety after receiving the NIPD results. A number of participants expressed the dilemma of wanting to know 'good news' but not wanting to know 'bad news'. Nevertheless, all the women interviewed still felt that they would rather know the sex of the fetus as soon as possible, even if this knowledge resulted in increased anxiety and difficult decisions having to be made, as it gave them time to prepare for what was ahead.

> 'It made you worry more, but then good worry because you had that knowledge, if you see what I mean.'
> Cathy – hemophilia carrier

Connection to a fetus when the pregnancy may be terminated. For a few of the participants who had opted for invasive testing and termination, a connection or tangible vision of a possible future appeared to have been made unintentionally as a result of knowing the sex.

> 'Knowing it was a boy, when I got to about ten or eleven weeks I was starting to feel very slightly, I looked at it differently, like 'oh, this is getting harder now."
> Jade – Pelizaeus Merzbacker disease carrier

This appeared to create additional stress and anxiety, particularly for those women that went on to have a termination of pregnancy.

Robbed of surprise. A number of women commented that knowing the sex took away the element of surprise. Nevertheless, this was perceived by most as a small price to pay.

> 'Knowing you've got a healthy baby to me is much more important than knowing what the sex is or you know, having that element of surprise.' Annabelle – AIS carrier

Misuse of technology. A few participants had concerns about whether NIPD might be used for social sexing purposes and there was a strong feeling that it would be 'unethical' to use this test in such a way. Furthermore, one participant raised the point concerning what the potential effect of future uses of NIPD might be on the disabled community.

DISCUSSION

Until now research has focused on attitudes of women and the public towards the availability of NIPD for chromosomal abnormalities and fetal sex determination.[17–19] Here, we evaluate the service users' direct experience of using NIPD for sex-linked genetic conditions and have identified an overwhelmingly positive attitude of participants towards this technology as well as a number of practical and psychological benefits of a test that is easy to perform, is offered relatively early in pregnancy and has no risk of miscarriage. A number of disadvantages were raised. However, participants felt these were fairly minor in comparison with the advantages. Further, some of the disadvantages raised, such as 'robbed of surprise', relate to any form of fetal sex determination.

In addition to the clinical benefits derived through NIPD including safety, ease of performing the test, early testing and reduction in the number of invasive tests being performed,[20] other psychological benefits were also identified. These included normalization of pregnancy, control and peace of mind. The absence of distress was also found which is a psychological benefit that sharply contrasts with women's experiences of invasive testing, which carries a risk of miscarriage.[21] Regarding the disadvantages of NIPD, very few issues were raised by participants and concerns such as the burden of 'unnecessary' decision-making for pregnancies that may spontaneously abort[22] were outweighed by the potential benefits of NIPD. One concern raised by a number of participants was around social sexing. This issue was also identified in a study looking at public attitudes towards this technology.[19] In the UK, NIPD for fetal sex determination is only used for clinical indications and this is tightly regulated by NHS service laboratories.

Three decisional pathways were identified in this study. An interesting finding related to the way in which some participants appeared to create distinct timeframes by using a stepwise approach to decision-making. This may have been a coping mechanism, which enabled them to retain a sense of control over the situation. A similar finding was reported by Scully *et al*[23] who describes women undergoing prenatal testing as 'narrowing the temporal depth of field of their attention' as a mental coping mechanism during a major life event.

The majority of carriers interviewed had made decisions around invasive testing before NIPD. Impact on reproduction has been highlighted as a key issue when learning about one's carrier status in other studies focusing on single-gene disorders,[24–27] and therefore this result is not surprising. These observations emphasize the need

for expert pre-test counseling before screening and add to concerns that implementing NIPD may erode informed choice.[28]

A recent study has identified variation in how services in UK offer prenatal testing options such as fetal sex determination and invasive testing to women who are carriers of hemophilia, with some services in the UK only offering NIPD to carriers of severe forms of the disease.[29] This probably relates to the fact that some health professionals consider that as gender could be identified at 20 weeks by routine scan, NIPD confers unnecessary extra cost.[29] However, here we have shown that there are often complex and dynamic issues such as personal experience and impact of other family members that are factored into the decision-making process, which were considered not only by carriers of severe forms of the disease. For these women, NIPD was still perceived to be valuable for both practical and psychological reasons, facilitating a sense of control and peace of mind early on in pregnancy. Thus, to ensure that NIPD is offered in a way that reflects patient need, it is important that we take into account the wider variety of benefits associated with NIPD. Moreover, NIPD should be offered in a standardized way to ensure equal access to all, alongside appropriate counseling and support.

Women undergoing invasive testing have been found to delay maternal attachment until the health of the fetus is confirmed.[30–33] This detachment or 'tentative pregnancy' is likely to be a psychological defense strategy enabling the woman to cope with the knowledge that the pregnancy might end in termination.[30] This phenomenon was observed in a number of women in this study for whom invasive testing was an option. It is possible that these women were delaying maternal attachment for this reason and might help us to understand why the experience of the ultrasound during invasive testing was distressing. Seeing an image of the fetus made the pregnancy more 'real', and hence it became difficult to remain emotionally detached.[34] Interestingly, in 'normal' circumstances, maternal attachment has been shown to increase significantly across time[32,35,36] with highest overall levels of attachment in the second trimester.[37] NIPD is therefore likely to be valuable for those receiving news in the first trimester that the fetus is of the unaffected sex as they are able to emotionally invest in their pregnancy before the time when the bonding process naturally peaks.

Increased anxiety was identified as an issue for those women who were still at elevated risk after receiving NIPD results. Nevertheless, these participants valued the information derived through NIPD even if it was news that the fetus could be affected, over the increased anxiety that resulted. The information appeared to give participants a sense of control and autonomy over their pregnancy, enabling them to make informed decisions around invasive testing and cope more effectively with their situation. For those who received news that the fetus was of the unaffected sex, NIPD was valuable as it provided relief from uncertainty early on in the pregnancy. Control, empowerment and relief from uncertainty have all been identified as patient-desired outcomes of genetic counseling[38–40] and therefore support the benefits of NIPD identified in this study. Furthermore, NIPD was found to enhance the decision-making process. This finding again supports what has been identified as a central goal of prenatal testing; facilitating optimal decision-making.[41]

Study limitations

The participants in this study were self-selecting, so the findings may be biased toward those that have strong positive views about NIPD. The study has not included carriers of sex-linked conditions that have chosen not to have NIPD, so we cannot comment on their viewpoint. Given the small number of participants (and the different subsets within the sample eg, condition), the findings cannot be said to be representative of the population. In addition, the majority of participants who took part in this study were white, hence the findings may not be transferable to underrepresented minorities. It would be interesting to explore the views of other ethnic minority groups as they may have different views and experiences of NIPD.

CONCLUSION

Service users had overwhelmingly positive opinions about using NIPD for fetal sex determination, with many more advantages than disadvantages being identified. Besides the practical advantages of earlier testing and avoiding miscarriage, there were a number of psychological advantages such as perceived control, normalization of pregnancy, peace of mind and facilitating decision-making. Until definitive genetic diagnosis using NIPD is available, NIPD for fetal sex determination is perceived as a good interim measure with a number of notable advantages over traditional methods. Further qualitative and quantitative research as and when NIPD becomes available for diagnostic purposes will be essential if we are to ensure it is offered to women and couples in a way that reflects their needs and preferences.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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