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ORIGINAL ARTICLE

The scope of prenatal diagnosis for women at increased risk for aneuploidies: views and preferences of professionals and potential users

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Abstract The increasing number of prenatal diagnostic tests in prenatal screening strategies, raises the question what tests to offer and why. This qualitative study investigated the views and preferences of professionals and potential users regarding four diagnostic test options for women at increased risk for common aneuploidies. Seven focus group sessions were conducted in The Netherlands between October 2009 and June 2010, with various categories of participants (n=55): professionals engaged in prenatal testing and potential users of this testing (meaning pregnant women and parents of young children). Participants were invited to mention all pros and cons and their preferences regarding four hypothetical diagnostic test options, presented on vignettes: a standard offer of rapid aneuploidy detection, karyotyping or array comparative genomic hybridization, representing a narrow, traditional and broad test, respectively, and the option of individualised choice. Then, a semi-structured group interview was conducted. The data were analysed by the constant comparative method. Participants identified similar test-specific pros and cons but showed different preferences. Users' opinion on what test to offer as a general policy differed from what they would choose themselves. All participants agreed that in theory, users should be enabled to make an informed choice about what test to apply, but they disagreed about the feasibility of this ideal. Standard narrow testing was favoured for its limiting effects on emotional and organisational burdens; individualised choice was preferred for assuring women's decisive influence. The varying opinions reflect different views on what autonomy in the prenatal screening context means, suggest that a single standard test offer is inadequate and that differentiation will be needed.

Keywords Prenatal screening · Views · Preferences · Qualitative research · Ethical issues · Reproductive choice

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Introduction

Current prenatal screening cascades include a first-trimester risk assessment for common trisomies and mid-trimester foetal anomaly scans. Invasive diagnostic testing is offered to women at an increased risk of chromosomal or genetic abnormalities (Salomon et al. 2011; Tapon 2010). Conventional karyotyping, that can reliably diagnose numeric chromosome aberrations and (major) structural abnormalities, has long been the standard diagnostic test. But alternative techniques, that allow testing for either a more

limited or broader scope, are increasingly being introduced instead. Techniques for rapid aneuploidy detection (RAD) target only trisomies 13, 18 and 21 and mostly sex chromosomal abnormalities (Faas et al. 2011). Other techniques, such as microarray-based comparative genomic hybridisation (array CGH), are used to detect a broader range of heterogeneous abnormalities (Hillman et al. 2011; Savage et al. 2011). Whereas some favour targeted arrays to detect selected abnormalities and avoid unclear or troublesome test results (ACOG The American College of Obstetricians and Gynecologists 2009; Shaffer et al. 2008; Van den Veyver et al. 2009), others prefer genome-wide, non-targeted arrays to maximally identify all kinds of clinically relevant aberrations (Choy et al. 2010 ; Faas et al. 2010). The implementation of these different techniques has led to debate about what diagnostic test should be offered to pregnant women and why (de Jong et al. 2011; Shuster 2007). There is no consensus among professionals about what specific test to offer in the prenatal testing cascade and pregnant women themselves have different preferences regarding the scope of testing as well (Boormans 2010; Bui et al. 2011).

Empirical studies on the preferences of professionals and 'users' (pregnant women, prospective parents and consumers) regarding the scope of prenatal testing have mainly focused on determining the differences with regard to choice outcomes amongst and between these two categories of respondents and on identifying the correlation between preferences on the one hand and characteristics of the tests and/or of the respondents on the other hand. Studies included a limited number of tests only. Boormans et al. (2009) and Grimshaw et al. (2003) introduced three test options: a test targeted on Down's syndrome only, RAD and karyotyping. These studies showed that testing for Down's syndrome only was hardly chosen, that professionals mostly wanted to offer RAD, and that opinions differed among pregnant women and between professionals and women. 'Individualised choice' was suggested as a solution for these differences (Boormans et al. 2009). Pieters et al. (2009) studied pregnant women's attitudes towards 'microarray-based genomic profiling technologies', which they referred to as 'prenatal full-scale testing for genetic disorders'. They found that this kind of test was not readily accepted, but that women with a low educational level were more likely to be interested in full-scale testing. More research into the factors that influence pregnant women's decisions was thought to be needed. When studying consumers' opinions toward reproductive testing, Hathaway et al. (2009) found a wish to have prenatal testing for more conditions than generally offered. These further conditions included for example mental retardation, deafness and cancer. The studies mentioned give a good overview of what test would be preferred by whom. We however wanted to know on the basis of what considerations professionals and users come to a certain choice. Therefore, we aimed to explore the spectrum of underlying views that play a part in the evaluation of prenatal tests and influence decision making. Given the current broad range of available techniques, we studied these views with regard to four prenatal test options: three diagnostic tests that are actually offered in current prenatal testing cascades: conventional karyotyping, a narrow test (RAD), and a broad test for heterogenous abnormalities (array CGH). Offering women an 'individualised choice' (Boormans et al. 2010) between these three tests was introduced as a fourth option. Although in current north-west European practice this option is limited to a choice between RAD and karyotyping (Boormans et al. 2010; Faas et al. 2011; Bui et al. 2011), we included array CGH into this alternative as well, because this test is increasingly applied. To explore the underlying views of professionals and potential users, we aimed to address three main questions: (1) how do Dutch professionals and potential users (meaning pregnant women and parents) evaluate the pros and cons of the four different test options; (2) what test options do they think should be offered to women in general and for what reasons; and (3) what test options do users prefer for themselves and why?

Materials and methods

Data collection

This study was undertaken in The Netherlands, between October 2009 and June 2010. Focus groups were conducted with potential users of prenatal testing and professionals who are involved in prenatal testing. We preferred a group-based approach over individual interviews, because groups are an effective source for eliciting views, for stimulating people to share and debate these, and for generating new ideas (Ulin et al. 2005). We composed homogenous groups, to effect that participants could speak freely among their peers and to reduce the risk that some participants would dominate the discussion based on their professional background or experience, or that others would feel uncomfortable or inhibited to contribute (Ulin et al. 2005; Polit and Beck 2004).

Informed consent was obtained at the beginning of each session. Users were offered a monetary incentive; all participants were offered a reimbursement for travel expenses. This study was cleared for approval by the Medical Ethics Committee of the Maastricht University Medical Centre+, azM, Maastricht, The Netherlands.

The focus groups were moderated by WD, ADJ or GDW and co-moderated by ADJ or JB. The group sessions all lasted 2 h. All focus groups started with a brief plenary introduction of the aim of the study and of the session, namely to explore all

views of participants and explicitly not to reach consensus. Then vignettes containing four test options were distributed (Box 1). The four options involved three standard test offers: (A) conventional karyotyping, (B) RAD and (C) a non-targeted array CGH. The fourth option (D) entailed asking pregnant women to choose themselves between (A), (B) and (C). We used vignettes to make sure that all participants had the same picture and a basic understanding of the four options. We described these test options in general terms and did not distinguish between different types of RAD or variants of array CGH. Instead, we focused on the common factors to avoid that participants, and especially professionals who actually offer these tests, would start to discuss the facts and details of different test techniques. The users and midwives had also been e-mailed this vignette the week before the group session, to make them already familiar with the subject to be discussed.

We sketched the scenario that women at increased risk for trisomies 13, 18 and 21 were offered a diagnostic test performed on material obtained by an invasive procedure (either amniocentesis or chorionic villus sampling). We chose this scenario because it resembles current practice that participants were likely to be familiar with, in order to avoid that it would be experienced as too hypothetical. After briefly explaining the content of the vignettes and answering informative questions, participants were asked to write down individually all pros and cons of the four test options and to give their opinion on what option should be offered to pregnant women. Users had to indicate as well what they would choose for themselves. Subsequently, they were asked to mention and explain the arguments and preferences they had written down. By following this procedure we wanted to ensure that all participants' initial thoughts were mentioned, and to prevent that the discussion would too soon strike out on a certain course. All arguments and preferences were listed on a flip-chart, to guarantee that they were all covered and rightly understood. To explore the considerations more in depth, a semi-structured group interview was conducted, guided by both a predetermined topic guide and the flip-chart notes. This qualitative design enabled to bring forward the individual views and to profit from the dynamics of a group discussion. All group sessions were digitally recorded and transcribed.

Box 1 Vignette with four test options

Option A – current
Pregnant women are offered a standard test: karyotyping
This test can identify all chromosomal abnormalities, including Down syndrome.
Some of these abnormalities have severe and others have mild clinical consequences
Generally, clear information can be provided about abnormalities found
Sometimes, the clinical consequences of abnormalities are unknown
Test results available in 2 – 3 weeks

Option B – narrow
Pregnant women are offered a standard test: RAD
This test can identify a few most severe chromosomal abnormalities, including Down syndrome.
Clear information can always be provided about abnormalities found
Test results available in 2 - 3 days

Option C – broad
Pregnant women are offered a standard test: array-CGH
This test can identify all chromosomal abnormalities, including Down syndrome
This test can also indentify other kinds of abnormalities, such as the hereditary disease cystic fibrosis
Often, clear information can be provided about abnormalities found
Sometimes, the clinical consequences of abnormalities are unknown
Test results available in 2 - 3 weeks

Option D – choice
Pregnant women are offered all three tests: A, B and C
They may choose themselves which test they prefer

Sampling and recruitment

As potential users we included pregnant women and parents (both male and female) of young children (0–3 years). The assumption was that these persons had been offered testing and would possibly have to deliberate about testing in a next pregnancy. We included fathers in the latter group, because of their partaking in the reproductive decision-making process (Locock and Alexander 2006; Williams et al. 2011). We included three categories of professionals, whose professional involvement in prenatal diagnostic testing differs: midwives have mainly initial involvement in counselling and reference to specialist care; gynaecologists from regional and urban hospitals are involved in the standard-test procedures; experts from academic centres actually perform specialised (follow-up) testing on a daily basis. By selecting these categories of participants, representing different perspectives, we aimed to collect a broad range of relevant views from persons directly concerned with prenatal testing (Polit and Beck 2004).

The users were recruited in the south-west of The Netherlands by advertisements in local newspapers and by distributing an information leaflet to pregnant women who visited the obstetric outpatient clinic at Leiden University Medical Centre. We recruited the midwives and academic experts from across the country by sending a digital general invitation trough their national professional societies. To reach relevant gynaecologists across the country, we asked the academic experts for possible referrals.

The sample

Seven focus groups were conducted that included 55 participants. Of these, 25 potential users were divided over four group sessions: two groups with in all 15 pregnant women (user pregnant=UP) and two groups with in all ten parents of young children (user male and female=UM and UF, respectively). We included 30 professionals who were divided per profession over three group sessions: one group with nine midwives (professional midwife-PM), one group with ten gynaecologists from regional and municipal hospitals (professional gynaecologist=PG), and one group with 11 experts from academic medical centres (professional academic=PA). The latter consisted of gynaecologists (n=4), clinical geneticists (n=3), cytogeneticists (n=2) and clinical cytogeneticists (n=2). Because in the course of the group sessions no new information was obtained and redundancy occurred, data saturation was achieved (Morse 2000) and no further participants were included.

Analysis

Analysis was conducted on the content of participants' notes, the flip-charts and the transcripts to identify major themes and sub-themes. The coding process started on the content of the notes and flip-charts, and followed the constant comparative method (Strauss and Corbin 1998). To reduce the chance of bias and ensure consistency, two members of the research team (ADJ and WD) independently coded the content systematically and iteratively, with guidance from an experienced qualitative researcher (AK). In this first phase of the analysis, we did a descriptive coding to label the various topics in the text. These open codes differed minimally and were adjusted slightly. The resulting scheme was used for subsequently analysing the text content of the transcripts, using the NVivo8® software program. Here, the same method was applied: ADJ coded the full content and WD checked for consistency by coding and comparing segments across the transcripts. In the second phase, we grouped the topics into sub-themes and found the same patterns in all three sources of data by means of an inductive reasoning method. The text belonging to these sub-themes were found to differ with regard to the 'level' of discussion: the test features as such were mentioned and classified, but the concepts of informed choice and autonomy elicited a more reflective discussion. This led to the distinction between the themes of 'evaluation of test features' and 'informed choice' as presented in the 'Result' and 'Discussion'. Participants' and users' preferences were approached as separate subjects, in order to give insight in how users differentiate between themselves and 'the public'. All researchers contributed to the discussion in the analysis process with regard to the formulation of the major conclusions. Thus, investigator triangulation was achieved.

Results

Themes and preferences

Two major themes emerged in the focus groups. Participants first focused on features of the test options, such as test outcomes, wait for results and costs and classified these as pros and cons. Secondly, a more reflective discussion started about the meaning of informed choice and autonomy in the prenatal screening context. The results caused us to distinguish between two groups of professionals, namely midwives and physicians (gynaecologists and experts from academic medical centres); we will refer to them accordingly. Pregnant women and parents together are called users. We talk about participants if we mean both professionals and users. The most important sub-themes are elaborated in the sections below and are often illustrated by representative quotes. Sometimes, citations are slightly adjusted for reasons of readability. Whenever an aspect relates to a specific test option, the denominator (A, B, C or D) is added between brackets. Participants' choices and reasons to choose a particular test option and users' preferences for themselves are presented subsequently.

Evaluation of test features

Test outcomes

Considerations regarding the amount and type of abnormalities possibly generated by the tests were mentioned by all participants. In general, more extensive information about the foetus was favoured. Users tended to associate broad testing (C) with certainty and narrow testing (B) with 'fake certainty'. UP13: '(…) test C (…) is the most comprehensive test, with complete clarity, because you know everything possible. (…) The disadvantage of B is that you don't have complete clarity and you can be falsely reassured as there may be other abnormalities'. Although professionals thought that more information could also 'falsely reassure' women, they stressed that a broad test could identify relevant abnormalities that would otherwise be missed. Yet, all realised that the profit of possibly generating more abnormalities would come at the price of an increase in (unclear) findings that would complicate counselling and decision making and aggravate the dilemma whether to terminate the pregnancy or not. A pregnant woman (UP2) commented: 'One has to take in a large amount of information and the spectrum of findings will become very large, so it will be really difficult to make a choice'. And a professional (PG2) stated: 'Some of the test results, such as deletions and translocations, are complicated and we hardly know what to do with them (…). It is not always clear what it all means to the foetus and this makes it difficult for the layman. And for us as well'. Some professionals mentioned that C could also generate unexpected findings about the parents and predispositions, which would be problematic if couples had not been adequately counselled beforehand. PA10: 'In addition to what has been said about C: an array does not only generate information about the foetus or causal mutations. It can indirectly also generate genetic information about the parents and about risk factors, such as a BRCA mutation (…)'.

Participants agreed that a narrow test (B) would avoid these problems while still identifying some severe abnormalities. Still, testing for all kinds of serious disorders (C) was found important. Users were particularly interested in hereditary diseases and limitation to some specific abnormalities (A and B) was debated. UP2: '(…) it started with Down's syndrome, because one could easily test this, when naturally there are other disorders that are even more serious. This is positive with C, that you can also find those other abnormalities and take them into account'. However, all recognised that differentiation between serious, mild and marginal abnormalities was difficult. Because of that, some participants feared that broader testing (C) would lead to the pursuit of a genetically perfect child and that abortion would increasingly be performed for minor abnormalities. UP5: 'Where does it stop? I think that the danger is that if there are possibilities to know more, that people increasingly will want to know more, and then it becomes the norm, a slippery slope. This frightens me'.

Finally, some found that less information would make the miscarriage risk of invasive testing disproportionate. PG3: 'It is dangerous to do an amniocentesis. So the moment that choices about the scope have to be made, my approach would be do it to the best of the possibilities available'.

These considerations about broad and narrow testing show that participants struggled to find the balance between their wish to test for all kinds of severe abnormalities and the drawbacks of too much findings.

Wait for results, costs and familiarity

Tests differ with regard to throughput time. Participants favoured quick test results (B) for three reasons. First, the period of stress and anxiety while waiting for test results would be minimised. PG5: 'I think that speed will reduce fear. There is a big difference between waiting for results for 3 weeks or 3 days'. Second, if test results were generated earlier in pregnancy, this would leave more time for the decision to terminate or continue the pregnancy. UP1: 'One might have more time to make that decision in peace and quiet. And surely with the second test (amniocentesis, ADJ) you have to decide rather quickly whether you want an abortion or not. This decision that you have to make within 2 to 3 weeks will be decisive for your life'. Third, earlier results would enable a possible earlier abortion, which was thought to be emotionally less traumatic. PM8: 'The pregnancy progresses too. So if women choose an abortion, then they of course rather have the termination of pregnancy at 16 weeks than at 18 weeks. Or at 9'.

The financial costs of the tests were approached from different perspectives. Most professionals asserted that costs, including time and personnel needed for counselling and logistics, would be high for C and D, and they considered this problematic from a public policy point of view. This problem was recognised by some users but 'solved' differently. They proposed public funding for a minimum set of tests (A and B) and (income related) out-of-pocket payment in case of more expensive testing (C and D) to ensure availability of all options. Other users, taking the individual user's perspective, found it unseemly to talk about financial costs of prenatal testing, because it would complicate the already precarious situation of prospective parents who were offered testing.

Finally, nearly all users appeared to only be superficially familiar with the organisation of current testing cascades and unaware of the fact that women are generally offered conventional karyotyping (A). This led to a striking contrast between professionals and users in how they evaluated A. While the longstanding experience with and reliability of A

was considered a great asset by professionals, users thought that this test did not have any advantage at all. UM2: 'A is neither here nor there. So if you have A, B, or C, than I would go for B or C. I think A is a compromise and combines the disadvantages only'.

The discussion about these aspects shows that only a short waiting time is unanimously favoured, but that participants differ in their evaluation of financial costs and testing experience.

Informed choice

Individualised choice: ideal and feasibility

In theory, all participants favoured an 'individualised choice' as presented in D. Terms mentioned were: promotion of autonomy, ideal informed choice, and control over own pregnancy. At the same time, they realised that the scenario presented in D would encounter practical drawbacks, because it would be difficult to adequately inform and counsel women and such a complex choice could overtax women and throw them off balance. UP10: 'With regard to D, the choices can be overwhelming and then you are not competent to make a rational assessment. So where do parents base their choices on, then?' Despite agreement on these both sides of D, participants reached opposite conclusions when discussing the feasibility of D in practice. Two groups could be discerned: (1) those who were sceptical about the feasibility of D, and (2) those in favour of D being dedicated to achieve it.

The first group, including most physicians and some users, considered it as good as impossible to provide women with adequate information and ensure adequate counselling. Especially physicians were sceptical about women's capacity to make the choice presented in D. PG4: 'It's a lot to ask of your patients. And the question is also if people are really waiting for all that information and all those choices. Mostly they'll be at a loss'. It was called a 'spurious option' (PG3), because it would provide women with a sense of control which would not correspond with the actual situation. PG7: 'And D, hmm, how is a layman going to make this choice. (…) except the advantage that men and woman have a feeling of control concerning their lives, there is no other real advantage in giving them the possibility to choose'. Users in this group stressed the emotional burden of choice and doubted whether women actually wanted to make it. UF4: 'I can imagine that as a pregnant woman you might find it scary to make that decision, and that it would be a lot easier if one test was simply offered'. Thus, the overall conclusion of this group was that D might provide for autonomous choice in theory but not in practice.

Participants in the second group, consisting of most midwives and most users, found that women's reproductive options should not be constrained by others. They considered the choice for a specific test to be an essential part of women's autonomy and especially midwives felt uncomfortable with an intrusion upon it. PM6: 'People have to make this choice themselves. Especially if various test alternatives are available, who do I think I am to withhold this information from them?' This view was placed in the societal context of an increasing individual freedom and availability of choices in many fields of personal life, including health care, and was reinforced by the awareness that women very much differed in their views and preferences. UP7: 'That you have a choice what type of test you would want, just gives you more freedom. Some want all available information even if the consequences are not clear, others do not. Personally, I would only want to know the information that is a hundred percent certain and if I can understand what it means. But that is me, I think that is different for each individual'. Still, there were ambivalent feelings toward the choice scenario. Midwives sensed that professionals devolved their responsibility (to choose the right test) unto pregnant women. PM1: 'You are putting the responsibility with the parents. But the feeling that you are passing the buck, makes me feel uncomfortable and reluctant'. The users in this group expressed worries regarding the responsibility and emotional impact of such a choice. They feared making the wrong choice and subsequent regret, particu-larly if the test chosen would miss an abnormality that would make the child suffer after birth. UP3: 'The moment that you have decided against these tests… can you forgive yourself if your child is born with an abnormality and dies after three pain-filled and dreadful years. How well can you look at yourself in the mirror with the knowledge that you could have known. There is no going back, that is what I find difficult about these tests'. The midwives and users in this group also thought that the complexity of such a choice could have the effect that women would (want to) rely more on professionals. Therefore, they emphasised that great effort should be put in informing and counselling women to enable them to make the choice offered in D. Since a standard approach would probably not meet women's individual needs, it was thought essential to tailor both the test offer and the counselling to women's different capacities and wishes. UF2: 'Just give the woman what she needs. I think that a 16-year-old pregnant girl will be frightened by all these options. But a 40-year-old woman is going to think more realistically, yes, there are risks. Well, I'm going to dwell upon all these options'.

These considerations show that the difficulties and possible drawbacks of individualised choice are acknowledged, but that this group wants and expects to overcome these with adequate counselling in order to respect women's autonomy.

Choice: consistency and fairness

Related to participants' different views on individualised choice itself, they dissented on how the testing cascade should be evaluated. The scenario in our study was that diagnostic testing was offered to women whose risk-assessment showed an increased risk for trisomies 13, 18 or 21. This led to discussion about the relation between the two successive tests. Those who were sceptical about C and D, took the risk-assessment as their point of departure and argued that the scope of subsequent diagnostic testing should be consistent with that (B). Possible additional findings of A and C were labelled as 'excessive' diagnostics by some physicians. PG2: 'In my opinion, it is an advantage that the results are the same as what the patients have undergone the screening for; that was the reason to do this. The results of the diagnosis would be equivalent to what you are screened for and nothing else'. Some users agreed that if the first test was for Down syndrome, one should not be troubled with avoidable other findings later on. Furthermore, it was thought 'unfair' if only women at increased could test for more abnormalities, while others, having the same risk for these surplus findings, had no access to this test.

The midwives and users who placed great importance on women's own choice, exactly reasoned the other way around and argued that access should be attuned to the favoured scope of diagnostic testing. They criticised the current focus on the trisomies and thought it unfair that women who were not at risk for these particular abnormalities had no access to broader testing. UP6: 'I don't think this is logical. I am forced to be tested for chromosome abnormalities for which you need to be at high risk and only after that am I allowed to test for hereditary disorders, whereas I might only want the foetus to be tested for hereditary abnormalities'. Therefore, they proposed to adjust the policy of admittance in order to enable access to broader testing for those women who wanted it.

Views and preferences

When asked what specific test options should be offered to pregnant women and why (research question 2), a majority of participants tended to choose between two extremes: either D or B (Table 1). Most users and midwives chose D, because it allowed women (and their partners) to make their own choice on what test would best suit their personal situation. Most physicians and the other users chose B for various reasons. Feasibility in terms of women's competence, counselling and costs, and consistency with women's indication (increased risk for trisomies) were mentioned most by the physicians. The emotional impact was decisive for users: B would only produce information about severe and clear abnormalities and therefore avoid difficult decision making and emotional burden. Only a few participants chose options in between: long-standing familiarity with A caused some professionals to chose this test, whereas C was seen as the best alternative between D (too complex) and A and B (too restricted).

Table 1 Test option that should be offered to women according to participants

Preference participant	A	B	C	D	No test
Pregnant women (n=15)		4		11	
Parents (n=10)		4	1	5	
Midwives (n=9)	1	1		6	1
Academics (n=11)	2	8		1	
Gynecologists (n=10)		9	1		

The picture changed when users indicated what test option they would prefer for themselves (research question 3) (Table 2). Their preferences were spread over four options: B, C, D or no testing at all. Reasons for B and D were the wish to limit the emotional burden of unclear findings and decision making, and to have the opportunity to make one's own choice, respectively. In this respect, there was no difference with the reasons mentioned for their 'general' preference. But some users who preferred D in general, did not want to bear the responsibility in this option themselves. The accumulation of decision-making moments throughout the pregnancy was experienced as an additional burden. UP8: 'The largest disadvantage for me would be that there has been an offer of choice in asking if the mother to be is willing to take part in the risk-assessment and in the high risk amniocentesis. And to make a further choice once more then between 3 different tests would be really difficult for me. So, one option would be enough for me'. Therefore, they chose the test that was most comprehensive (C) but still not too burdensome.

Finally, part of the users indicated that they would choose no testing at all, either because they were opposed to abortion or would not want to risk losing the pregnancy because of iatrogenic miscarriage, whatever the possible benefit of testing.

Discussion

This qualitative study on professionals' and users' views and preferences regarding four prenatal test options showed

Table 2 Test option that users prefer for themselves

Preference participant	A	B	C	D	No test
Pregnant women (n=15)		4	5	6	
Parents (n=10)		3	1	1	5

two levels of deliberation: classification of test features and reflection on the concepts of informed choice and autonomy. The latter seemed to have the largest influence on participants' preferences. Furthermore, personal bearing capacity led some users to give a different answer with regard to what they would choose for themselves and to what choices should be offered to pregnant women in general.

Features of testing: pros and cons

Initial observations regarded the features of tests, including test outcomes, wait for results and financial costs. Since other studies have shown the relevance of these 'test attributes' (Boormans 2010) in the evaluation of prenatal diagnostic tests (Boormans et al. 2009; Grimshaw et al. 2003; Pieters et al. 2009), we expected these issues to arise in our focus groups as well. Users and professionals identified similar test-specific aspects, but their evaluation did not coincide in all respects. The same inventory of aspects however suggests that users in our study realised both the beneficial and adverse aspects of various tests—despite existing doubts whether they have adequate understanding of the drawbacks of particularly broad testing (Pieters et al. 2009). Although users understood that, compared with karyotyping (A) and RAD (B), more and different abnormalities could be identified by broad testing (C), it was not clear whether they realised that the incidence of these other findings is considerably lower. Remarkably, this lower incidence was not a major issue for the professionals either when evaluating the various test options. However, pre-test information should include a distinction between common findings and possible but rare findings (Wertz et al. 2003) and may differ in both quantity and detail. But the question remains how to provide complex information in the actual counselling situation without confronting users with either a deficit or an overload of information. In either case users would not be adequately equipped to make an informed choice to either participate or not engage in testing. Moreover, user's individual comprehension and the time–pressure in the prenatal context should be taken into account. Although we observed that users and professionals identified the same set of test-specific features, these appeared not to be decisive for choosing a specific test option.

Reproductive autonomy: limiting burdens or maximising options

Although all participants agreed that reproductive autonomy was in theory best served by leaving the decision for a specific test to pregnant women themselves, the feasibility of such an informed choice in practice was evaluated differently.

The view that an informed choice as intended in option D and needed in option C is too complicated and that optimal decision making and limitation of burdens should prevail over maximising information, may be seen as paternalistic (Beauchamp and Childress 2009; Dworkin 1988). However, adherents of a narrow test seemed to assume that limiting choices better accords with the aim of prenatal screening to enable autonomous reproductive choice, given the requirements of, amongst other things, adequate counselling, informed consent and proportionality (benefits for participants outweigh the burdens) (Health Council of the Netherlands 2008). This view also leaves room for what we have earlier called the 'logic of the screening strategy' argument (de Jong et al. 2009), holding that the limited scope of the preceding risk-assessment justifies and requires offering a corresponding diagnostic test to ensure consistency throughout the testing phases. In this view, a broader test is not rejected per se but may be offered for specific indications. Indeed, broader testing (C) has already been introduced and was favoured by professionals in our study in case of foetal ultrasound abnormalities (Faas et al. 2010; Savage et al. 2011). Although it seems only logical that problems regarding comprehension and counselling will also arise in this situation—albeit on a smaller scale—these were hardly discussed for this context. In this regard, the justification for limiting the scope of testing and reproductive options was contextualised: physicians tended to reason from an organisational point of view, taking the existing screening system as their point of departure.

Participants preferring D reasoned primarily from a users' perspective. They asserted that reproductive autonomy required enabling an individualised choice: allowing pregnant women themselves to determine whether they would benefit more from a comprehensive or a limited test. In this view, the scope of and access to diagnostic testing should not be limited by others but valuable options should be available to all. Still, the possible disadvantages of such a choice was acknowledged. Users' fear for making a wrong decision and regret afterwards—the so-called anticipated decision regret (Tymstra 2007)—and midwives' concern about professionals' failure to take their responsibility reflect the anticipation of a so-called decisional conflict. Such a conflict may arise when people are faced with an inherent difficult choice and other complicating factors (O'Connor et al. 2002) such as—in this case—lack of knowledge and time pressure. To obviate these drawbacks, extensive counselling was thought to be crucial.

Although counselling models as such were not discussed, it was obvious that participants thought that a situation in which professionals only provide 'objective' information and subsequently leave the decision making to the testee alone, would have serious shortcomings. This so-called information model (Emanuel and Emanuel 1992) has

formerly been criticised for being too simplistic and inaccurate, for the misconception that a strict distinction between facts and values can be achieved, and for ignoring professionals' duty to help patients handle information in order to reach an autonomous choice (Emanuel and Emanuel 1992; García et al. 2009; Marteau and Dormandy 2001). Precisely this latter aspect of responsibility to support is well addressed in the 'interpretive model', in which the professional not only provides the patient with adequate information but also acts as a counsellor who engages in patient's autonomous decision making. The interaction between professional and patient thus aims to 'elucidate the patient's values and what he or she actually wants and help the patient select the available medical interventions that realise these values' (Emanuel and Emanuel 1992). Because the agenda of the user is directive in this process, the interpretive model may also meet the ambivalent feelings that users expressed with regard to extensive counselling: that women presumably want to be supported by professional guidance when facing a difficult choice, that such support can make women depend on and be influenced by professionals, which may both contrast with autonomous choice. If the counselee herself determines the direction and content of counselling, these possible drawbacks can be countered. This would also enable to meet the different interpretations that women may have of informed choice and the specific support they expect from the professional (Ahmed et al. 2012). Nevertheless, in view of the doubts expressed about the feasibility and desirability of more extensive counselling, there is a need for further ethical reflection and empirical research on what counselling model would be appropriate in prenatal testing scenarios to ensure that women are really enabled to make an autonomous reproductive choice.

Users' preferences: general policy versus private choice

A majority of the users expressed similar considerations and preferences for themselves as for women in general. However, a large minority mentioned new arguments and different preferences when deliberating about what option they would choose for themselves as opposed to what should be offered to women in general.

First, some declined all forms of testing because either abortion as such or the procedure-induced miscarriage risk was unacceptable to them. Importantly, these users emphasised that they did not want to impose their personal choice on others: every woman should make up her own mind what option best suited her personal situation and whether she would terminate a pregnancy or run the risk of losing a healthy foetus. Therefore, they still approved of the general offer of testing. As for their personal preferences, a correlation between declining prenatal testing and abortion is consistent with other studies (Lumley et al. 2006; Potter et al. 2008). The iatrogenic miscarriage risk is also a known reason for rejecting prenatal testing (Caughey et al. 2008). It is likely but as yet unclear whether the introduction of non-invasive prenatal testing (Chiu et al. 2011) means that these particular users will accept testing if the miscarriage risk is absent or at least the number of unnecessary invasive procedures will be reduced (Bianchi et al. 2012). If so, the question remains if they would test for either a narrow or broad range of conditions, as soon as the latter becomes possible (Lo et al. 2010; Fan et al. 2012).

Second, half of the users who appreciated D in general chose C for themselves. This alternative of a standard broad test enables maximising information, albeit at the cost of possible difficult test-outcomes and decision making, but avoids the burden and responsibility to choose the 'right test'. This dilemma between wanting a choice and experiencing ambivalence about subsequent decision making in the prenatal setting has been described before (Aune and Möller 2012) and reflects awareness of the chance of a decisional conflict. These considerations reinforce the need for giving adequate support to individual women in their decision-making process.

Concluding remarks

The controversy shown by the views and preferences in our study reflects one of the central ethical questions in current prenatal screening, namely how reproductive autonomy in this context has to be interpreted. Does it mean that the amount of reproductive options for women should be maximised and individualised choice be offered to all? Or should the burdens be limited, the decision-making process be optimised and a standard narrow test be offered in order to ensure a real autonomous choice? Our results confirm the finding in other studies that the question what test option best complies with the aim of prenatal screening is answered differently by both professionals and users, and suggests that a 'one-size fits-all' approach fails to take into account the different views and preferences of pregnant women and their partners (Kupperman and Norton 2005). Since both a broad and a small standard test offer may interfere with women's autonomy, differentiation may be needed. The ethical and practical conditions for such a differentiated test offer are still to be defined. Whatever the test offer may include, our findings suggest that not only adequate information and non-directivity are crucial in counselling, but that there is also a need for reflection on counselees' values and for support to realise these. The interpretive model may be helpful in this regard.

Limitations

This study has several limitations. First, the sample size was limited and our sampling method cannot rule out that participants might have been biased and may not be representative of the Dutch professionals and users. This means that findings cannot be generalised without caution. Second, in this study participants reflected on a hypothetical situation and persons' hypothetical responses may not match actual behaviour. These limitations did however not hamper the aim of this study to explore views and preferences of potential users and professionals. The general criticism of qualitative research, that it is too subjective and that researchers' beliefs contribute too much to the findings (Mays and Pope 1995; Press 2005), we refute as follows: the data collected were rich in content as all participants could among their peers express their views and give comprehensive explanations, and the analysis was conducted independently by various researchers. The strength of this study is that various categories of participants, who can be considered stakeholders of prenatal testing policies, were enrolled and that they provided a broad range of considerations that can inform further reflection on the scope of prenatal screening.

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