

Making Use of NIPT: Rethinking Issues of Routinization and Pressure

Abstract

Mapping the main ethical issues surrounding prenatal testing, we then analyze which concerns are specific to non-invasive methods. Presupposing the *privatization premise* for reproductive autonomy in fundamentally liberal societies, we go on to specify common concerns about NIPT covered by the term ‘routinization’, and conceptually unravel the frequently expressed worry of increasing ‘pressure’ to test and/or terminate affected pregnancies. We argue that mindful decision-making should be a key educational goal (not only) of NIPT counseling which could be achieved through stepwise disclosure. In addition, we identify indirect social pressure as the most plausible threat to reproductive freedom. While continuous efforts need to be made to prevent such pressure – not least by ensuring balanced availability of options –, restricting testing options, and thus freedom of choice, cannot be the answer to this concern. Lastly, we suggest abandoning the vague term ‘routinization’ and instead focusing on specified concerns to enable a fruitful debate.

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Introduction

Non-invasive prenatal testing (NIPT) is a recently introduced method that allows to identify certain genetic characteristics of a fetus. Currently, these include but are not limited to chromosomal aberrations. It is to be expected that sooner rather than later medical-technological progress will render a variety of other uses possible. [1]

Undeniably, in terms of medical benefit and safety NIPT is entirely advantageous over other available prenatal diagnostic tests (PND) such as amnio- or choriocentesis: While these invasive interventions involve a small risk of procedure-induced miscarriage, NIPT requires nothing but a blood sample from the pregnant woman and therefore poses no risk to her pregnancy. Due to its non-invasive nature, it is physically and mentally much less burdensome. Moreover, it can be conducted earlier in pregnancy and more discretely. It is, however, just NIPT's procedural harmlessness or triviality that in the eyes of critics gives rise to some specific ethical concerns of which the fears of "routinization" and "pressure" seem the most prominent rhetoric coatings.

Mapping Ethical Concerns Regarding NIPT

Objections to the provision and the use of NIPT are usually not directed against the test *itself*, but against its role as a promoter of selective abortions. Notwithstanding some uneasiness with the term 'selection' especially in German contexts [cf. 2], it has become the internationally established label for abortions motivated by certain

characteristics of the potential off-spring – e. g. by genetic abnormalities. Selective abortions, much as abortions in general, are viewed as ethically illegitimate by those who ascribe a right to life to human embryos. In addition, ending pregnancies in order to avoid giving birth to a disabled child is often criticized because of these acts' (alleged) discriminatory implications or consequences for living people with disabilities. And finally, ethical concerns are raised, that selective reproduction might on a general scale deteriorate parent-child-relations or de-humanize society. In the absence of empirical data that would give evidence to such undesired social developments in the wake of PND and lacking a consensus on the moral status of early embryos, secular societies tend to be more or less liberal towards selective abortions and thus to respect private autonomous decision-making by individual women and couples on these very personal and moral matters. Let us call this the *privatization premise*, which in this paper we will take for granted and, moreover, for appropriate and legitimate.

Against this background, any plausible objection against NIPT would have to question the *autonomy* of its usage. Thus, the two ethical objections that *specifically* address consequences of NIPT's procedural triviality, have to be looked at from this perspective. Critics claim or worry that NIPT might:

- turn into a *routine* test;
- go along with some external *pressure* on women to actually make use of this easy-going test. Both physicians and women's social environment, so the concern, might more and more *expect* the use of NIPT, slowly turning it into a *de facto* screening tool.

Both concerns somehow allude to violations of the autonomy requirement for valid informed consent. But do these concerns survive scrutiny?

In their perennially influential monography *Principles of Biomedical Ethics*, Beauchamp and Childress develop a model of decision autonomy with three necessary preconditions, i.e. patient “competence”, sufficient “understanding” and the absence of “controlling influence” by others – influence that would render consent in-voluntary. Thus, to count as valid, any patient authorization of a medical intervention has to meet these three requirements [3, chap. 4]. Issues of defective or questionable competence are typically related to consent in the context of psychiatric or pediatric care or else with regard to patients suffering from compromised cognitive, emotional, or volitive capacities. For the clientele of pregnant women and their partners, this aspect seems of marginal importance for single cases only. Obviously then, the above two concerns against NIPT would, if justified, regard the understanding- or the voluntariness-requirement of NIPT consent.

Specifying ‘Routinization’

As reported above, NIPT’s benefits are often accused of contributing to its ‘routinization’. Indeed, this concern has been at the center of the debate from its very beginning [see for example 4, 5]. Although not normative in itself, in the context of NIPT this term is mostly used with a decidedly negative connotation. But a clear-cut definition is often missing.

This lack of conceptual clarity has only recently received more attention: ‘Routinization’ has been identified as an umbrella term [6] or a container concept [7] that encompasses a number of underlying concerns. Profiting from their insights, we want to add to the picture by starting from a more general definition.

According to the Oxford English Dictionary (OED), ‘routinization’ means “the fact of being or becoming routine in character or operation” [8]. With an eye to this entry, we will concentrate on ‘routine in character’, since operative excellence cannot as such reasonably be criticized in any (medical) context. In turn, the OED defines ‘routine’ as

[...] a regularly followed procedure; an established or prescribed way of doing something; a more or less mechanical or unvarying way of performing certain actions or duties [9].

Following the OED, we suggest differentiating between four meanings of NIPT becoming “routine in character”, each of which we want to evaluate from the perspective of liberal ethics, i. e. presupposing the *privatization premise*, as spelt out in the first section:

(i) NIPT will quite likely be used with increasing frequency and – by not only substituting, but amending PND – lead to a rise in overall prenatal testing. But, as has been stated before, an increasing usage *as such* need not be problematic, as long as it does not occur for problematic reasons [6]. Assuming that women opt for NIPT in the very informed and thoughtful manner that is strived for in all prenatal testing, nothing could and should be criticized – in accord with the privatization premise (see

above). Hence, one must take a closer look at the decision-making processes that might lead to women's increased utilization of NIPT. In short, it is not the pure quantity, but the quality of choosing NIPT that might turn out ethically problematic and might even occur unnoticed. Again following the above OED definition, three such qualitative transformations seem conceivable:

(ii) The decision for NIPT following a 'routine' could mean to take an "established" course of action. 'Established' is a neutral term but can be interpreted in different ways. It could mean, for instance, that NIPT becomes more widely accepted (as expressed by individual autonomous decisions). This in turn could have an alleviating impact on women who would want to use prenatal testing, but whose social environment is currently intolerant of this decision. These women might be more confident in daring to choose NIPT in accordance with their personal liberal convictions if their decision is an 'established' one. To illustrate this by an innocent (and less ethics-laden) example: Some mothers might have wanted to take their young children to kindergarten but could not do this against conservative familial convictions. Routinization of early onset external child-care might make it easier for them to follow their own preferences.

(iii) Critics of NIPT view it as a subtly "prescribed way of doing something", as included in the OED definition. More often, such suspected 'prescription' is called 'social pressure' by which NIPT would slowly become the social default – so that women might no longer dare to decline the test. We will look at this frequently expressed worry further down. In any case, pressure could potentially impair the voluntariness condition of women's decisions about NIPT.

(iv) Finally, women might decide in favor of NIPT in “a more or less mechanical or unvarying way”. This could be interpreted as a lack of mindfulness or of deliberative diligence regarding the potential personal and moral consequences of a positive test result. Opting for “just another blood test” [10, 11] without realizing its specific meaning for potential future decisional burdens, might indeed violate the understanding condition of autonomous decision making. We will come to this right away.

Understanding

According to the standard view of informed consent, its validity requires quite diverse categories of knowledge on the side of the consenting person. Above all, she must understand that she individually has the unlimited right to authorize or veto any intervention at stake. In addition, she ought to be informed about the intervention’s nature, its comparative risks and benefits, and its potential consequences in any relevant regard. The relevance in question should ultimately be judged from a personal perspective.

In the context of prenatal genetic testing – invasive or non-invasive – ‘pathological’ outcomes (for example a detected chromosomal aberration) inescapably confront pregnant women with the option of deciding for or against continuing a thus affected pregnancy. While some women would not even want to receive genetic information about their future child, others are interested in knowing about their future child’s potential disabilities in order to prepare for a life together. So far however, a majority of women or couples opt for testing because they intend or would consider terminating

their pregnancies in case of an abnormal test result. And in case of such a result most of them do decide in favor of an abortion. [12]

As has been emphasized by various authors, autonomous decision-making both before testing and afterwards, requires complex knowledge about testing, its possible results, its safety and reliability, and about the effects that genetic risks of disease or disability could have on the future child and its family. Moreover, women should be informed about potential decisional burdens and the non-medical nature of selective decisions: Despite prenatal genetic testing taking place in a clinical setting, the decision to terminate or to continue an affected pregnancy is (outside rare vital problems on the side of the woman or the fetus) not primarily medical in nature, but deeply impregnated by personal interests and moral views. When confronted with a deviant test result, women should get detailed information about financial, social, and medical support in caring for a child born with the genetic deviation at hand. They should be enabled to connect with parents who are or have been in similar situations. All of this being true for prenatal genetic testing in general, what's specific about NIPT? Some authors fear that autonomous decision-making about NIPT might require an "information overload" [13]; others are concerned that the procedure's very triviality might reduce the attention given to careful and broad pre-test disclosure on the side of physicians as well as of test-using women [14]. Seeing the risk of disclosure overload as a general rather than an NIPT-specific issue, we do, however, embrace the second concern. Thus, in agreement with many other authors, not the least a number of Dutch colleagues, we find it very convincing to frame NIPT by a well-structured setting of stepwise disclosure [15]. For example, this could include a general information offer and, in case of a positive reaction, an exploration of which genetic characteristics of

the fetus the pregnant woman would be interested in. Only after this personal scope is set, the desired concrete information would be conveyed. This approach could prevent inattentive or insufficient counseling as well as issues of information overload. It would therefore be useful not only in the context of NIPT, but for prenatal testing in general. Even if empirical data on informational deficits in NIPT might still be inconclusive, there is much to say in favor of preventing any such deficits to occur or aggravate. In particular, counseling experts should neither assume sufficient ex-ante knowledge on the side of their patients nor completely postpone any discussion on the potential termination of the pregnancy until the occurrence of a deviant test result.

Conceptualizing Pressure

Increasing 'pressure' upon women is a predominant issue of critical concern related to offering and using NIPT. As analyzed above, it is one of the core suspicions framed as 'routinization'. Once again, however, the term 'pressure' is often used without sufficient definition or characterization. While some authors in their analyses presuppose a common understanding of "undue pressure" [14], others apply it to a variety of situations, ranging from "explicit coercion on the part of healthcare providers to subtle influences of prevailing social norms" [16]. In what follows, we want to shed some conceptual light on this key term and draw some normative conclusions.

According to the standard view in medical ethics stipulated above, decisional autonomy requires, among others, "freedom from controlling conditions" [3, chap. 4]. They conceptualize this "voluntariness" condition mainly as the absence of coercion and undue manipulation by other persons or institutions on the decision at stake.

Outright coercion being obviously irrelevant for the context of NIPT in a fundamentally liberal society, manipulation seems to be the phenomenon to look at for further orientation. Here, Beauchamp/Childress focus on “informational manipulation” as “a deliberate act of managing information that alters a person’s understanding of a situation and motivates him or her to do what the agent of influence intends” [ibid, p. 137]. Unsurprisingly, they consider many forms of such informational manipulation as “incompatible with autonomous decision making. For example, lying, withholding information, and exaggeration with the intent to lead persons to believe what is false” [ibid]. On the other hand, they caution against an inflated diagnosis of control by manipulation in health care:

We often make decisions in a context of competing influences, such as personal desires, familial constraints, legal obligations, and institutional pressures, but these influences usually do not control decisions to a morally worrisome degree [ibid, p. 138].

Where does this leave us with regard to NIPT decision making? First of all, it can never be completely ruled out that individual counselors or caretakers intentionally exercise informational manipulation in order to nudge or even direct women into undergoing/not undergoing a prenatal test or a subsequent abortion [17]. But on the whole and in a transparent society like ours, it is very unlikely for such unprofessional behavior to occur on a larger scale and to remain undetected and uncriticized. Nevertheless, binding professionals to the standard of non-directive counselling during their training and continued education seems both advisable and feasible.

Yet another form of pressure that critics are afraid of is the slow conversion of NIPT as an option for single cases into a screening tool whose use is widely regarded as part of a responsible and reasonable reproductive care regime. On the other hand, not using NIPT could then gradually be seen as careless, irresponsible, and even objectionable. To be sure, raising and supporting disabled children often require additional financial, emotional, or organizational efforts by families as well as by society in terms of educational or health care support. Circumventing such perceived 'burdens' might, so a common worry, slowly invite subtle pressure towards NIPT and subsequent selective abortions. By no means does this seem a totally ungrounded speculation.

In theory, three varieties of such a potential development could be distinguished. In the first scenario pressure towards NIPT and selective abortions might result from society's gradual direct or indirect reduction of support and inclusion of disabled children. In a liberal and non-discriminatory society this would be a constitutional, ethical, and political no-go, which would have to be banned in any respect. As a social prognosis it cannot be precluded in principle but does not deserve serious concern in today's modern societies. Moreover, empirical data prove a significant increase in positive attitudes towards the inclusion of disabled children [cf. 18].

In the second scenario pregnant women might not adequately realize the complex personal and moral issues at stake. Or they might not adequately perceive of their unconditional rights to both reproductive autonomy and social support in raising a disabled child. The underlying mechanism would thus start with poor knowledge

leading into a spiral of slowly shifting the default line in prenatal testing habits. Quite obviously, this coincides with the already discussed violation of the understanding condition. As a result, future pregnant women might un mindfully adapt to ‘what everybody does’. We emphasize once more that such deficits in understanding should by all means be prevented.

In a third scenario, women would well know about their rights to reproductive autonomy (including the use of NIPT). But despite being thus well-informed, they might *feel* under pressure to adapt to an increased use of NIPT resulting from autonomous decisions by other women. This possibility touches some deeper issues of freedom in pluralistic societies, which we now want to address. According to social philosopher Philipp Pettit, freedom of choice presupposes the *availability* of options (“open doors”) regardless of one’s own preferences and regardless of what others prefer the agent to choose [19]. Again, like doors, options can be more or less easily accessed. Thus, intentionally rendering options difficult to take, is yet another way to limit someone’s freedom.

Basically, NIPT issues involve option-freedom. Women should thus be well informed about the availability of the test and about their freedom to use it or not to use it – without having to justify their decision to authorities and without having to overcome obstacles that are meant to remind them of the socially unwanted character of either option. Trivially, having more options available for one’s reproductive behavior might entail decisional burdens, but that is the price to be paid for freedom – here as

elsewhere. Protecting women from choice-overload by restricting the availability of options would surely be a paternalistic no-go.

All that having been said, how should one assess the reverse concern of the third scenario above – i. e. the concern that women might *feel* pushed to use the seemingly comfortable option-door of NIPT? It has indeed been a long-standing argument in the pressure debate that the invasiveness and riskiness of PID so far has functioned as a firewall welcomed by some women who repudiate prenatal testing and selecting for moral or principled personal reasons [20]. With NIPT they would now lose their defensive “pretext” against society’s potentially increasing pro-test expectations [13].

Surely, withholding medically advantageous options from women to protect them from having to explain their decision-leading moral or personal reasons cannot be an adequate solution for preventing potentially contested or heteronomous decisions. Such a solution must rather focus on the way NIPT is framed and portrayed. Procedural aspects must be adjusted to empower and encourage women to make autonomous, individual decisions independent from perceived expectations.

On the other hand, the options of not-testing and thus, if the situation occurs, of accepting and raising a disabled child must become a still easier option (a wide ‘open door’) than it often is in reality. Social pressure consisting in unintended but nevertheless effective neglect of the various needs of affected women, children, and families must be confronted and combatted. Such invisible indirect social pressure seems to be the greatest danger for reproductive freedom – much as in other contexts where options are asymmetric in terms of societal costs and efforts. An analogous

example might be seen in end-of-life decisional options: Critics of liberal regulations of assisted suicide regularly express worries that over time society might diminish its efforts in making life satisfying even under conditions of terminal disease, frailty or advanced dementia. The required antidot is sometimes called “equally valuable options” [21] or making available options “real” ones [22]. We prefer subsuming these ideas under the expression ‘balanced availability of options’ as this might even better illustrate the need for symmetrically open doors.

Once more then, our answer to this problem points to the preconditions of autonomous decision-making: without some stamina, some maturity, some willingness to take final responsibility for how to lead one’s life, the whole idea of autonomy collapses. Literacy in reproductive medicine and reproductive ethics has to be among the educative goals that should to some extent be realized even before women and couples come to procreate. Utmost emphasis should be put on the *privatization premise* (see above) implying that neither societal disapproval nor social approval should significantly alter reproductive options and decisions.

Talking of ‘societal’ influences somewhat vaguely refers to impacts by health care institutions, counselors, or public climate. In contrast, personalized influences, for example, “perspectives of partners, family members and friends” are not commonly believed to fall into the category of ‘pressure’ but are rather seen as unproblematic “social context[s]” [23]. Needless to say that here, too, repressive or irresistible influences can violate reproductive autonomy – yet another issue that needs mentioning in attentive reproductive education and counseling.

Practical and Normative Conclusions

Throughout this paper we have presupposed the *procreative privatization premise*, according to which women and couples should be free to decide upon contraception, abortion and prenatal testing according to their own personal and moral convictions. For NIPT policies, five consequences would in our eyes have to be drawn from this premise.

Firstly, NIPT should in principle be available for every pregnant woman (at most at a small fee – topic that we cannot address here).

Secondly, stepwise professional counseling [7, 15] should be standard practice to prevent understanding deficits as one potential reason for non-autonomous decision making. Mindful deliberation must be seen as a common goal of counselee and counselor.

Thirdly, society must very carefully monitor the balanced and easy availability of options for couples facing the risk or the certainty of giving birth to a disabled child. Indirect unintended social pressure towards the less challenging and costly option of selective abortions should be realized as a real danger to both the autonomy of women and the human liberalism of society.

Fourthly, reproductive literacy vis-à-vis complex option-sets has to rank high on the agenda of public education – already before people's actual procreation.

Fifthly, the vague worry of 'routinization' should be abandoned in favor of the more specified concerns above. Routinization rhetoric has become a common framing for ethical uneasiness with liberal positions in bioethics. It should neither be inflated nor obscure those issues that deserve serious debate and action.

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