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SCIENCE AND SOCIETY

Advances in prenatal screening: the ethical dimension

Antina de Jong, Wybo J. Dondorp, Suzanna G. M. Frints, Christine E. M. de Die-Smulders and Guido M. W. R. de Wert

Abstract | Prenatal screening strategies are undergoing rapid changes owing to the introduction of new testing techniques. The overall tendency is towards broadening the scope of prenatal testing through increasingly sensitive ultrasound scans and genome-wide molecular tests. In addition, non-invasive prenatal diagnosis is likely to be introduced in the near future. These developments raise important ethical questions concerning meaningful reproductive choice, the autonomy rights of future children, equity of access and the proportionality of testing.

Current prenatal screening cascades in the developed (primarily Western) world consist of similar testing methods^{1,2}. These include a first-trimester risk assessment for the most common aneuploidies and a second-trimester fetal ultrasound scan to identify fetal anomalies³. In the case of a positive risk assessment, invasive procedures (amniocentesis or chorionic villus sampling) are offered to allow diagnostic follow-up testing. For decades, conventional karyotyping has been the gold standard for this form of prenatal diagnosis. However, it is now being replaced by molecular techniques that will allow narrowing or broadening of the scope of diagnostic testing (FIG. 1). This raises questions about the range of conditions for which such testing should be offered and why. A distinct development is the pending introduction of non-invasive prenatal diagnosis (NIPD). This promises to make prenatal screening easier and safe but again raises the question about its scope. As we discuss here, there is an important ethical dimension to the developments and choices that the field is currently facing.

Most of the ethical questions discussed in this paper are relevant for shaping future prenatal screening practices both in settings where services are largely offered as a market-driven commodity (as in the United

States) and in countries where they are part of public health care (as in most European countries, Canada and Australia). Standard-setting and debate about guiding principles have contributed to the formation of an internationally acknowledged normative framework for the provision of prenatal screening (BOX 1). On a general level, this framework builds on the basic principles of biomedical ethics: respect for persons, beneficence, non-maleficence and justice. More specific norms for population screening include proportionality, equity of access and informed consent⁴. A core element of the normative framework for prenatal screening is that informed consent as a mere precondition is not enough. Enabling meaningful reproductive choice with regard to parenting or avoiding a child with a serious disorder or disability is (or should be) the very aim of offering testing for fetal abnormalities^{5,6}. This is in order to ensure that abortion decisions remain personal and are not turned into instruments of societal goals, such as prevention and cost-reduction by bringing down the number of people requiring life-long and costly care.

Here we provide an overview of current developments in prenatal screening, showing how each of these relates to the normative framework and what ethical challenges they entail. We then address some ethical issues

in more detail and make suggestions about how specific challenges might be addressed. Although we take the current normative framework as our point of departure, our message in this paper is that the dynamics of the field may also put the framework to the test.

Because selective abortion is a major issue of moral sensitivity in debates about prenatal screening, it seems appropriate to make the following preliminary remarks. A first debate concerns the moral status of the fetus. The prevailing view, also reflected in legislation allowing abortion up to a certain limit, is that the status of the fetus is relatively low and increases with its development. Those who take this view usually accept that abortion may be justified if the parents want to avoid having a child with a serious disease or disability⁷. Although it is sometimes proposed that society should come up with a list of diseases serious enough to warrant prenatal testing, it is widely accepted that this is unrealistic and undesirable, given that many disorders have a variable expression and that the impact of a disease is, to a large extent, context-dependent⁸. A second debate concerns the selective nature of abortion for fetal abnormalities. According to the 'disability rights critique', prenatal screening sends the message that it would have been better if those living with the targeted conditions had not been born⁹. However, the claim that this is true by implication does not hold. Whether prenatal screening has discriminatory effects depends on how the programme is presented and implemented in practice. If informed decisions rather than abortion rates are taken as a measure of success, the disability rights critique is less convincing⁷.

A final remark on terminology: we use the term prenatal screening to refer to the whole of the testing trajectory offered to low-risk pregnant women. Prenatal diagnosis refers to follow-up testing as a part of this trajectory. Prenatal testing is used as a generic term.

Developments in prenatal screening

Rapid aneuploidy detection. In most developed countries, the diagnostic test after a positive risk assessment for trisomies 21, 18 and 13 is still conventional karyotyping^{1,10}.

Figure 1 | Prenatal screening cascade: current and possible future test routes. Test routes in current and possible future prenatal screening cascades. The horizontal arrow at the top denotes the gestational age. In the case of an increased risk of trisomies 21, 18 or 13, invasive prenatal diagnosis is offered: conventional karyotyping, rapid aneuploidy detection (RAD) or genome-wide molecular testing. A nuchal translucency ≥ 3.5 mm or fetal ultrasound anomalies may lead to either follow-up ultrasound testing or invasive prenatal diagnosis.

Non-invasive prenatal diagnosis (NIPD) using cell-free fetal DNA or RNA that is obtained from maternal blood (plasma) may, in the near future, be implemented clinically as a one-step diagnostic test for trisomy 21 (and possibly also trisomies 13 and 18) using RAD or genome-wide molecular testing, including next-generation sequencing, for the detection of paternal mutations that underlie monogenic disorders. NIPD could also be offered as a follow-up test after ultrasound anomalies. See TABLE 1 for more details about each type of test.

This method detects all microscopically visible chromosome aberrations, including rare numerical or structural abnormalities that may have serious clinical consequences but also deletions, duplications or other chromosomal anomalies that can have mild, uncertain or even no clinical consequences^{11,12}. As a result, women may be confronted with other findings than those for which they were offered risk assessment and follow-up testing. This is one of the reasons why, particularly in northern European countries, some have advocated to replace conventional karyotyping with a targeted molecular 'rapid aneuploidy detection' (RAD) test^{13,14}. Three common RAD techniques are currently being used: interphase fluorescence *in situ* hybridization (FISH),

quantitative fluorescent PCR (QF-PCR) and multiplex ligation-dependent probe amplification (MLPA)¹⁵ (TABLE 1). These tests generate reliable results within 3 days, which is much faster than karyotyping, and are used to diagnose only trisomies 21, 18 and 13, although probes often also include the sex chromosomes. In terms of the normative framework, the question arises whether avoiding additional findings that may complicate decision making render RAD a better approach to diagnostic follow-up testing than karyotyping¹⁶. Or is the opposite the case, given that RAD reduces the range of possible outcomes that women may find relevant for reproductive choice¹⁷? Does the miscarriage risk attached to current invasive procedures require that testing should be

maximally informative¹⁷? While this debate is still going on¹⁸, both karyotyping and RAD are currently offered, either as stand-alone tests or as options between which pregnant women may choose¹⁹⁻²¹.

Prenatal ultrasound. Whereas the framework for prenatal screening was developed at a time when a limited number of fetal abnormalities were screened for, the introduction of fetal ultrasound screening has effectively changed this. The second-trimester fetal ultrasound scan has a wide range of possible outcomes. Apart from various structural defects (with severe, mild or unknown clinical consequences), these screens increasingly reveal so-called 'soft markers' that indicate a risk of a wide range of genetic disorders,

which require further diagnosis to confirm. Although 'controversial' soft markers may sometimes be discarded^{22,23}, the trend seems to be towards more detailed screening³. Remarkably, the question of how women can be properly prepared and give informed consent for this complex form of prenatal screening has, until now, received only scant attention²⁴.

Nuchal translucency measurement at around 11 weeks of gestation was introduced as part of the risk assessment for common trisomies but has developed into a *de facto* screening test for other defects as well. An enlarged nuchal translucency of ≥ 3.5 mm is associated with an increased risk of intra-uterine fetal death, congenital heart defects and many other structural defects and genetic syndromes. This finding generally leads to follow-up diagnostic testing by using further ultrasound scans, karyotyping or genome-wide molecular tests^{25,26}. However, pre-test information does not usually cover this possible further trajectory. This means that women may be confronted with outcomes they were not prepared for and may find difficult to handle.

Genome-wide molecular testing. The debate about RAD can be expected soon to give way to the question of whether the scope of diagnostic testing should be broader than karyotyping. That, at least, seems to be the thrust of technological developments²⁷. Molecular genetic tests with a higher resolution than conventional karyotyping include several array-based platforms and shotgun or parallel sequencing that can be used for genome-wide analysis^{29,30} (and perhaps, in the future, even whole-genome analysis²⁸) (TABLE 1).

Abnormalities that such tests might detect include small interstitial or telomeric deletions or duplications and partial or uniparental disomies that may, in addition to identifying childhood-onset disorders, also lead to the detection of genetic risk factors for late-onset disorders such as hereditary cancers. In some centres in Europe and the United States, genome-wide tests are offered to women at an increased risk of chromosomal abnormalities or in cases of ultrasound anomalies^{31,32}. Some propose targeted testing by using arrays that are designed to detect clinically significant abnormalities while minimizing the detection of unclear findings^{32,33}. However, others favour wider arrays in order to identify more aberrations³⁴. Although more comprehensive testing may have benefits in terms of clinically relevant findings that would otherwise be missed, a serious challenge is that,

with the present state of knowledge, many of the results of such testing are impossible to interpret. This leads to a lack of clarity about what women should be told and, more generally, to concerns about the feasibility of well-informed decision making for comprehensive testing. For example, copy number variants (CNVs) that are detected by array comparative genome hybridization (array-CGH) or SNP analysis can be categorized as likely to be pathogenic, likely to be benign or of unknown clinical significance³⁵ (TABLE 1). The balance of benefits and burdens will also be affected if unnecessary anxiety is caused by such outcomes. Further questions relating to genome-wide molecular testing are whether the autonomy rights of future children may be violated and whether justice would require offering such testing to all pregnant women.

Non-invasive prenatal diagnosis. In the next 2 to 5 years, it is expected that NIPD performed on cell-free fetal DNA (cffDNA) and cffRNA in maternal blood will be introduced as a routine screening test for aneuploidies (BOX 2; FIG. 1). In high-risk families, NIPD will also possibly serve as a routine diagnostic test for autosomal-dominant and autosomal-recessive traits, such as Huntington's disease and cystic fibrosis, respectively^{36–38}. In the short-term, this may be limited to the

detection of paternally inherited traits owing to difficulties in distinguishing between maternal and fetal DNA.

As the benefits of an early and safe one-step approach to prenatal diagnosis are clear, it would seem that current risk-assessment practices for common trisomies can be abandoned in the screening context. However, if nuchal translucency measurement — which is currently a part of this risk assessment — were also to be dropped, a much smaller range of abnormalities will be identified. Introducing NIPD may thus lead to a reduction in the scope for autonomous reproductive decision making by pregnant women. Although this loss can be averted by offering stand-alone nuchal translucency measurement as a complement to NIPD, that would make the testing trajectory more costly, complicated and burdensome. In terms of the aim of prenatal screening, piling up additional tests may have the adverse effect of undermining autonomous choice.

Some commentators expect that, eventually, genome-wide NIPD testing will become possible³⁹. Although this would reduce the need for complementary tests, concerns about the feasibility of adequate informed consent and decision making are heightened when broad genetic testing takes the form of a safe and simple blood test to be performed

Box 1 | Key ethics terminology

The 'normative framework' regarding prenatal screening for fetal anomalies refers to the set of ethical (and/or legal) norms that together provide the context of justification for professional practice and public policy making with regard to prenatal screening. This framework can be regarded as a context-specific articulation of the basic principles of health-care ethics⁴:

- The principle of respect for persons reflects the notion that, as autonomous agents, human beings deserve to be treated as moral subjects. In the context of screening, this requires that decision making about participation and all further steps in the screening process must be based on adequate information and voluntary choice ('informed consent').
- The twin principles of beneficence and non-maleficence stem from the Hippocratic roots of medical ethics. As screening is offered on the initiative of the health-care system, it is essential that the possible benefits for participants clearly outweigh the disadvantages (burdens and risks) that such programmes always also have — for instance, as a consequence of false-positive or false-negative outcomes.
- The principle of justice requires that equity of access to screening programmes is ensured and that they are provided on the basis of a fair distribution of benefits, burdens and financial resources.

Building on these ethical norms, the framework for prenatal screening fits in with the more general set of principles for population screening, as initially formulated by Wilson and Jungner⁴⁹. But prenatal screening has a different aim than most other forms of population screening^{6,50}. Instead of providing options for treatment or prevention of disease, prenatal screening aims at providing pregnant women and their partners with information that they may want to use as a basis for decision making about whether to continue the pregnancy. The 'clinical utility' of prenatal screening depends on the extent to which it allows participants to make well-informed and autonomous reproductive choices. However, (prenatal) screening programmes must also be proportional. This cannot be decided merely in terms of the balance of benefits and drawbacks for participants. One should also look at possible consequences for other individuals and groups and to societal implications, including costs⁵¹.

as early, perhaps, as 7 weeks of pregnancy⁴⁰. As NIPD will take away the 'gatekeeper' effect of the current risk of miscarriage that comes with amniocentesis and chorionic villus sampling, concerns have also been raised that it will contribute to testing for minor abnormalities or non-health related conditions and to a corresponding 'trivialization' of abortion³⁸.

Specific ethical issues

Meaningful reproductive choice. Given that prenatal screening should be aimed at providing pregnant women with opportunities for meaningful reproductive choice, a crucial issue is whether new tests expand or undermine these opportunities¹⁸. The classic interpretation of informed consent is that a decision to be tested or not must be

well-informed for every condition included in the test panel. Ideally, the woman undergoing the testing should also be given the opportunity to indicate which specific outcomes she does not want to be informed about ('the right not to know'). But the clear tendency towards ever-broader testing makes this model appear increasingly out of touch with reality. Broad prenatal testing may generate findings that vary from severe conditions, such as Duchenne muscular dystrophy, to predispositions for a wide range of early- or late-onset diseases and to CNVs for which the clinical consequences are unclear. To give detailed pre-test information on the level of all such outcomes is not realistic. It would lead to 'information overload' and would frustrate rather than serve the aim of autonomous choice⁷.

How can this problem be solved? One option is to look for an alternative approach to informed consent, avoiding information overload but still allowing well-informed decision making. A possible model proposed in the context of multiplex neonatal screening is that of 'generic consent' (REF. 41). This involves presenting pre-test information in general categories of types of outcomes⁷. In the context of prenatal testing, relevant categories include: congenital lethal disorders; early- or late-onset disorders requiring intensive medical care; early- or late-onset disorders requiring limited medical care; susceptibilities for complex disorders; conditions involving only minor health problems; and abnormal findings of which the clinical implications are unknown.

Table 1 | Summary of prenatal tests

Test	Test principle or technology	Test material used		Analysis	
		Non-invasively obtained*	Invasively obtained†	Targeted	Genome-wide
Fetal ultrasound scan	Ultrasonographic imaging of growth and anatomy	Not applicable	Not applicable	No	No
Maternal serum screening	Risk calculation based on biochemical quantification of proteins in maternal blood	Maternal serum	Not applicable	Yes	No
Conventional karyotyping	Microscopic analysis of chromosome banding patterns from fetal cells	Not applicable	Cultured fetal cells	No	Yes
FISH	Microscopic analysis of fluorescent labelled probes hybridized to specific genomic loci in interphase or metaphase fetal nuclei	Fetal erythroblast	Uncultured or cultured fetal cells	Yes	No
RAD [§] -FISH	Microscopic FISH analysis for aneuploidy (21,18,13) of fetal interphase nuclei	Not applicable	Uncultured fetal cells	Yes	No
RAD [§] -QF-PCR	Size-ratio (2/1) analysis or peak count of multiplex quantitative fluorescence PCR product using polymorphic short tandem repeats	cffDNA or cffRNA	Fetal cell DNA	Yes	No
RAD [§] -MLPA	Multiplex quantitative PCR-product ratio analysis using oligonucleotide probe pairs hybridizing to specific genomic loci compared to normal control	cffDNA or cffRNA	Fetal cell DNA	Yes	No
Array-CGH	Genome-wide mapping of chromosomal gains or losses (~CNV) by semi-quantitative ratio analysis of patient to reference DNA, simultaneously hybridized to substrate-immobilized probes	Not applicable	Fetal cell DNA	Yes	Yes
SNP array	Genome-wide mapping of chromosomal gains and losses (~CNV) or loss of heterozygosity by semi-quantitative analysis of patient DNA hybridized to substrate-immobilized oligonucleotide probes specific for single nucleotide polymorphic loci	cffDNA or cffRNA	Fetal cell DNA or RNA	Yes	Yes
Shotgun sequencing	Sanger (re)sequencing of small indiscriminate fragments of a whole genome	cffDNA or cffRNA	Fetal cell DNA or RNA	Yes	Yes
MPS	Whole-genome (re)sequencing by simultaneously sequencing hundreds of millions of immobilized short-sequence tags attached to a support	cffDNA or cffRNA	Fetal cell DNA or RNA	Yes	Yes
Exome sequencing	MPS of exon sequences only preceded by a genome-wide exon-capture step	cffDNA or cffRNA	Fetal cell DNA or RNA	Yes	Yes
Mutation detection	Gene-specific Sanger sequencing or customized mutation-specific PCR	cffDNA or cffRNA	Fetal cell DNA or RNA	Yes	No

*No risk for the fetus or unborn child (includes non-invasive prenatal diagnosis (NIPD)). †Includes chorionic villus sampling, amniocentesis and percutaneous umbilical cord blood sampling. §RAD of common aneuploidies on chromosomes 21, 18, 13, X and Y in 1 to 4 days. Array-CGH, array comparative genomic hybridization; cff, cell-free fetal; CNV, copy number variant; FISH, fluorescent *in situ* hybridization; MLPA, multiplex ligation-dependent probe amplification; MPS, massively parallel (next-generation) sequencing; QF-PCR, quantitative fluorescent PCR; RAD, rapid aneuploidy detection.

A key question is whether this still facilitates well-informed decision making. One problem is that the different categories may fail to capture the variable expression of many disorders. Moreover, the severity of different diseases is perceived differently among both health-care professionals and pregnant women. The practical feasibility of such alternative models of informed consent needs to be tested in empirical studies.

A second option is the use of filters to minimize findings that may be unclear or otherwise difficult to handle. This would allow counselling and decision making to be limited either to a few selected serious conditions or (in the context of broader testing and linked with a form of 'generic consent') to a smaller or wider subset of the six categories mentioned above. This approach has the advantage of avoiding the problem of whether pregnant women's interests are really served by being offered the choice to receive information about all possible outcomes of genome-wide molecular testing. If, as we think, the answer to this is 'no', it is inevitable that professionals and other stakeholders must find a balance between allowing women to decide for themselves as much as possible and helping them to do so by defining the range of meaningful choices.

The autonomy rights of the future child. A second issue that arises in the scenario of genome-wide molecular testing is that of the future child's 'anticipatory autonomy rights' (REF. 42). These rights may be violated when prenatal testing has revealed a predisposition for a late-onset disease that is not amenable to treatment or prevention starting in childhood and the woman decides to continue the pregnancy. Testing children for such conditions is generally regarded as unacceptable, not just because benefits for the child that might outweigh any burdens are absent but also because it deprives the individual of his or her right to self-determination. This includes determining whether to be tested once mature enough to do so^{43,44}. But then, clearly, it is also problematic to perform such tests in fetuses that may well grow into children. There are two possible solutions here. One solution is to limit the scope of testing by using targeted arrays or algorithms, avoiding as much as possible the detection of predispositions for late-onset diseases. The drawback of this is that women who might have chosen to terminate the pregnancy in case of such a finding are effectively deprived of what for them would be a meaningful reproductive option. The alternative solution is to allow

Box 2 | Non-invasive prenatal diagnosis

In 1997, Lo *et al.*⁵² discovered circulating cell-free fetal DNA (cffDNA) and cffRNA in maternal plasma, creating possibilities for non-invasive prenatal diagnosis (NIPD). The main advantage of this is that the procedure-related miscarriages of current invasive techniques (namely, chorionic villus sampling and amniocentesis) are avoided, as NIPD requires only blood samples, which are taken from the mother as well as the father. The cffDNA and cffRNA appear in the maternal blood circulation from early placenta formation (from 5 to 6 weeks of gestation — the trophoblast stage), which makes reliable NIPD possible from 9 weeks of gestation. NIPD is already implemented in clinical practice for fetal Y-chromosome detection in case of X-linked disorders and for fetal Rhesus D (RhD) determination⁵³.

Many research groups are now trying to detect the most common trisomies, 21, 18 and 13, through NIPD. These trisomies can be detected either by shotgun sequencing or massively parallel genomic sequencing^{54,55}. Recently, the first large-scale validation study for trisomy 21 has been published⁵⁶, and clinical implementation is expected within 2 to 5 years. Targeted NIPD for combined testing of trisomies 21, 18 and 13 remains more challenging. The number of known trophoblast-expressed genes is still limited, and as long as the use of epigenetic signatures that offer potential for detecting trisomies — for example, DNA methylation — has not become routine, it is difficult to develop a targeted, robust and cheap aneuploidy test^{57,58}.

women to undergo genome-wide analysis but only after they have expressed, in pre-test counselling, the clear intention to choose abortion if a predisposition for a late-onset disease is found. Although women cannot, of course, be forced to stick to this intention, this approach may provide the optimal balance of respecting the autonomy rights of the woman and those of her possible future child²⁸. The same approach is used in the context of prenatal diagnosis for Huntington's disease⁴⁵. Empirical studies are needed to see whether this would also be feasible in the different context of prenatal screening.

Equity of access. What about the justice of offering genome-wide prenatal tests only to women with a positive risk assessment for common trisomies? Other pregnant women may rightly complain that they have the same *a priori* risk for all other outcomes of such tests. This issue already arises with karyotyping as a diagnostic follow-up, but it becomes more prominent with genome-wide molecular testing, in which more abnormalities may be found and the detection/miscarriage ratio will be more favourable. This problem of formal justice can be theoretically eliminated in two ways. One is to only offer RAD as a follow-up test. The other is to offer invasive testing to all pregnant women, as is the case in the United States¹⁰. The former solution amounts to a form of 'levelling down justice': denying possible benefits to some because others cannot have them. Problems of capacity aside, the latter solution (access for all) seems to be most in line with the general aim of offering opportunities for autonomous reproductive choice^{18,46}. Although concerns about exposing women to the

risk of miscarriage without good reason are certainly relevant for counselling, denying access to invasive procedures on these grounds is paternalistic. This seems difficult to justify, given that the relevant tests may reveal serious fetal abnormalities. Of course, it does not follow that testing for whatever conditions women find important should be covered by public or collective funding. This is an issue of distributive justice and proportionality that we will briefly comment upon in the next section.

Overall proportionality. A crucial element in the moral evaluation of screening programmes is their proportionality, meaning that the moral importance of what the programme will achieve must outweigh any disadvantages. On the level of the benefits and burdens for individual women and their partners, the developments outlined above raise the question of how the tendency towards broader screening will affect this balance. To answer this question, data are needed about the psychosocial aspects and decision-making dynamics of wide-scope prenatal testing, including the possible implications for family members. As follows from our discussion of the expected introduction of NIPD-based prenatal screening, proportionality may also be challenged when new and better tests do not replace what was offered before but instead lead to additional testing. Moreover, the consequences of new developments for different social groups and society at large should also be taken into account. Testing for only some abnormalities (as is the case in using RAD) may reinforce the criticism that prenatal screening contains the message that certain groups of people are not welcome in society⁹. However, this

argument loses much of its sting with the further broadening of the scope of prenatal testing⁴⁷. If prenatal screening is used for a broad range of conditions, no groups need to find themselves specifically targeted.

On a societal level, the proportionality of prenatal screening is also a matter of cost-effectiveness⁴⁸. With ever-broader testing, this becomes more complicated than when prenatal screening was still directed at a few serious abnormalities, such as Down's syndrome — the costs of which for society are high, and the detection of which leads to, in most cases, a termination of pregnancy. It seems reasonable that public funding must cover prenatal testing for disorders that have a considerable impact on the lives of children and families but not necessarily more than that. Inevitably, this will refuel the debate about which conditions are serious enough to warrant prenatal testing, although it will do so from the different perspective of access and funding. Indeed, this may well lead to a shift towards giving more emphasis to considerations that are relevant from a public health perspective.

Concluding remarks

We have discussed four developments that challenge the current normative framework for prenatal screening. Our conclusion regarding RAD is that offering women the choice between conventional karyotyping and narrower testing is most in line with the aim of prenatal screening. With regard to ultrasound screening and genome-wide diagnostic testing (based on current invasive procedures or in the context of NIPD), we think there is a clear need to explore the feasibility of alternative models of informed consent. As it seems inevitable that filters will have to be used, debate is needed about how these should be defined. Genome-wide prenatal testing will have the effect of blurring the distinction between prenatal testing and predictive testing of minors. Until now, these were different worlds with their own normative frameworks, focusing, respectively, on reproductive choice for pregnant women and on the interests (including future autonomy rights) of the child. We have argued that, in the light of these interests, the right of the pregnant woman to obtain information about the genetic make-up of her future child is not unlimited, nor unconditional. Finally, the clear tendency towards ever-broader testing in the context of prenatal screening raises morally relevant questions about access, funding and proportionality. As this article has shown, the choices are as much about ethics as they are about technology.

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Competing interests statement

The authors declare no competing financial interests.

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