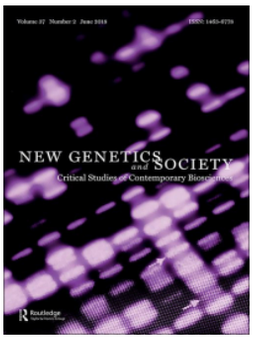

Routledge
Taylor & Francis Group

New Genetics and Society

Critical Studies of Contemporary Biosciences

ISSN: (Print) (Online) Journal homepage: www.tandfonline.com/journals/cngs20

The salience of genomic information to reproductive autonomy: Australian healthcare professionals' views on a changing prenatal testing landscape

Kerryn Drysdale, J. L. Scully, L. Kint, K.-J. Laginha, J. Hodgson, I. Holmes, K. L. MacKay & A. J. Newson

To cite this article: Kerryn Drysdale, J. L. Scully, L. Kint, K.-J. Laginha, J. Hodgson, I. Holmes, K. L. MacKay & A. J. Newson (2024) The salience of genomic information to reproductive autonomy: Australian healthcare professionals' views on a changing prenatal testing landscape, New Genetics and Society, 43:1, e2332310, DOI: 10.1080/14636778.2024.2332310

To link to this article: https://doi.org/10.1080/14636778.2024.2332310

Published online: 22 Apr 2024.

New Genetics and Society
2024, Vol. 43, No. 1, e2332310 (25 pages)
https://doi.org/10.1080/14636778.2024.2332310

The salience of genomic information to reproductive autonomy: Australian healthcare professionals' views on a changing prenatal testing landscape

Kerryn Drysdale[a], J. L. Scully[b], L. Kint[c], K.-J. Laginha[d], J. Hodgson[e], I. Holmes[d], K. L. MacKay[d] and A. J. Newson[d]*

[a]*Centre for Social Research in Health, University of New South Wales, Sydney, Australia;* [b]*Disability Innovation Institute, University of New South Wales, Sydney, Australia;* [c]*Sydney Medical School, Faculty of Medicine and Health, University of Sydney, Sydney, Australia;* [d]*Sydney Health Ethics, Sydney School of Public Health, Faculty of Medicine and Health, The University of Sydney, Sydney, Australia;* [e]*Department of Paediatrics, Melbourne Medical School, The University of Melbourne, Melbourne, Australia.*

(*Received 9 October 2023; final version received 12 March 2024*)

Genomic testing in prenatal care is rapidly advancing and it is now possible to obtain an entire fetal genome via a blood test administered in early pregnancy. In the pursuit of reproductive autonomy, more tests are being offered to more people, for an ever-increasing range of indications. Health professionals who provide pregnancy care are at the vanguard of prenatal testing, yet their views on the impact of technology advancements remain under-explored. Qualitative interviews with Australian healthcare professionals revealed that they value prenatal testing for its information-giving potential. However, the rationale underpinning testing reveals a tension between professionals' desire for certainty and patients' expectation that they will be reassured. Coupled with the greater salience of genomic information over other forms of knowledge in prenatal care, we suggest that the unchecked pursuit of information by way of genomic technologies in prenatal testing may paradoxically constrain reproductive autonomy in practice.

Keywords: Prenatal diagnosis; bioethics; genomics

Introduction

Caring for those who are pregnant or contemplating pregnancy almost ubiquitously involves discussions about prenatal testing. The rationale for offering such tests is to provide information about genetic or chromosomal anomalies that may inform decisions in the pregnancy, such as to continue or terminate.

*Corresponding author. Email: ainsley.newson@sydney.edu.au; @biomedethics

Prenatal tests were originally developed to offer those at "high risk" (for example, due to maternal age) information about three chromosomal aneuploidies: trisomy 21, trisomy 13, and trisomy 18 (Farrell 2013; Perrot and Horn 2022; Richardson and Ormond 2018; Schmitz 2013). Early forms of testing measured biochemical analytes in maternal blood, and so offered only indirect and probabilistic information about these conditions. Subsequent confirmatory testing required an invasive procedure that posed a risk of miscarriage (Löwy 2017).

Advances in both prenatal sample generation (such as non-invasive prenatal testing, or NIPT) and DNA sequencing (including next generation sequencing methods) now enable the generation of more accurate information about a fetus, while also posing low or no risk to a pregnancy (Van den Veyver 2016). These tests, enabled by technological innovation, offer greater reliability in the information available to healthcare professionals and the advice they can give their patients (Holloway *et al*. 2022). While still an advanced screening technology, NIPT has been rapidly taken up globally (e.g. Nature 2019; Pös, Budiš, and Szemes 2019; Ravitsky *et al*. 2021), including in Australia (Hui and Halliday 2023). Previous recommendations for risk stratification of testing have been superseded by recommendations of routine offers of NIPT as a first-line test (e.g. ACOG & SMFM 2020). In Australia, NIPT is predominantly offered through primary care, with specialist follow-up if results suggest an increased chance of the fetus having a genetic or chromosomal anomaly (McKinn *et al*. 2022). Alongside NIPT, tests such as microarray following diagnostic sampling (i.e., by way of amniocentesis) are widely available provided that certain clinical indicators are met. Further developments in testing, such as exome or whole genome sequencing of fetal DNA following diagnostic sampling, are being assessed under research protocols.

Alongside technological developments, the indications for prenatal genomic testing have expanded (Richardson and Ormond 2018; Schmitz and Henn 2022). More tests are being offered to more pregnant people, for more conditions. These tests are identifying genomic information of greater volume, complexity, granularity and unknown significance (Hashiloni-Dolev, Nov-Klaiman, and Raz 2019; Nature 2019; Navon and Thomas 2021). These increased offers and uptake of genomic information in pregnancy have numerous drivers, including consumer interest (e.g. Bowman-Smart *et al*. 2019), a fear of liability on the part of clinicians (Murdoch *et al*. 2017), and commercial imperatives (Baldus 2023; Kaposy 2017; Löwy 2022).

However, the burgeoning use of genomic tests in pregnancy remains contested (Christiaens, Chitty, and Langlois 2021; Dondorp, Page-Christiaens, and de Wert 2016; Donley, Hull, and Berkman 2012; Kaposy 2017; 2023; Navon and Thomas 2021; Schmitz and Henn 2022). For example, Navon and Thomas (2021) argue that expanded uses of genomics in prenatal testing are driven more by the availability of sequencing technology than by a consensus on indications. Ongoing concerns about utility are also reflected in test nomenclature: non-invasive testing was

at first "diagnosis," then "testing," and then (or also) "screening" (Ravitsky *et al*. 2021).

One important aspect of the debate over expanded prenatal testing is that genomic information remains an overall poor predictor of human health (Best *et al*. 2018; Richardson and Ormond 2018). The relationship between a genotype (a person's genetic constitution) and phenotype (their observable characteristics), and then how that phenotype is experienced as a life, can be hard to quantify. Even well characterized conditions such as Trisomy 21 (Down syndrome) have a variable phenotype (Bull 2020). Expanding the scope or indications for prenatal testing to a wider range of conditions will amplify the range of both phenotypic variability and how this influences one's lived experience (Baldus 2023). So, while next generation sequencing and non-invasive methods of accessing fetal genetic material can discern a DNA sequence or ascertain the presence or absence of relevant chromosomes, they cannot show what that sequence means for a future child's health or well-being (de Jong *et al*. 2014; de Melo-Martín 2006).

Concerns have also been raised about the technical limitations of tests like NIPT, including the possibility of more false positive results and low positive predictive value, especially for conditions of lower prevalence (McKinn *et al*. 2022; Wise 2022). The complexity of the genomic data that is now becoming more available will also continue to pose, and likely amplify, challenges to the embodied knowledge held by pregnant women (Bernhardt *et al*. 2013; Kringle-Baer 2017; Lippman 1999; Werner-Lin *et al*. 2017). An increased chance or high chance result following a test like NIPT does not mean that a fetus will necessarily be affected (Baldus 2023). Barbara Katz Rothman's ground-breaking Tentative Pregnancy (1986) highlights how information from emerging techniques confront pregnant people with new burdens and responsibilities, including how these techniques remove barriers to decline additional information through testing (p.82). Despite advances in medicine, the core dilemmas surrounding autonomy persist, indicating that "knowledge does not always empower." (p.176). Indeed, Rothman's early sociological critique of the novel uncertainties caused by prenatal testing seems even more relevant today.

The promotion or achievement of reproductive autonomy is fundamental to the justification of an expanded offer of genetic or genomic information in pregnancy. At a high level, reproductive autonomy can be considered as a person's ability to "reflect critically upon … preferences, desires and values" in reproduction and "to develop attitudes towards them" (Hildt 2002). Autonomy in general is widely considered as a good, and so by extension is reproductive autonomy. There is a now substantial literature specifically on what reproductive autonomy is and how it can be achieved (e.g. Jackson 2001; McLeod 2002).

Reproductive autonomy has been used as a grounds to justify expanded offers of genetic or genomic information in pregnancy (Benn and Chapman 2016; Donchin 1996). However, critics have raised the suspicion that beyond a certain point,

information provision in prenatal testing may not be autonomy-enhancing and may even be autonomy-impairing (Ravitsky, Rousseau, and Laberge 2017; Seavilleklein 2009; Werner-Lin, Mccoyd, and Bernhardt 2019). Focusing on the informational properties of a prenatal test distracts from the basic rationale for testing, and neglects the wider social and structural contexts in which decisions will be made (Donchin 2001; Gabriel 2017; Suter 2018). Because genomic information has a high degree of significance in the current context of prenatal care, it is increasingly given higher priority over other kinds of information about the pregnancy. On a more practical level, a health professional relaying genomic information may not do so in a way that is resonant to the pregnant woman (Hunt, de Voogd, and Castañeda 2005).

In providing pregnancy care, healthcare professionals have a significant role in influencing how emerging genomic technologies are offered and implemented, how the data they provide are interpreted, how information is presented, and how these features shape subsequent decisions. This means that they also provide (at least in part) the necessary conditions to facilitate reproductive autonomy (Hodgson and Spriggs 2005). However, there has been little research to ascertain what healthcare professionals think about the expansion of prenatal testing and its impact on reproductive autonomy. This paper addresses that gap. As one part of a larger mixed-method empirical bioethics project, we report findings from a qualitative study that sought to capture insights from healthcare professionals on what they understand reproductive autonomy to be, and how it is facilitated or inhibited in the context of increased offers of genomic information in pregnancy.

A note on terminology and our intended audience. Regarding terminology, we have used the terms "patient," "pregnant woman," and "pregnant person" throughout this article. Our aim is to adopt inclusive language, and to reflect how participants in this study used these terms, as well as "couple," interchangeably, often within the same interview. Our use of a variety of terms is intended to be inclusive without undermining the lived experiences of anyone who is pregnant or has a child. In respect of our intended audience, as a multi- and inter-disciplinary research group we hope that our paper presents and discusses our findings and the issues they generate to be accessible to those from a variety of fields. This would include those who may be able to translate our findings when caring for pregnant people themselves.

Materials and methods

This qualitative interview study forms part of a larger project that aims to refine and improve the concept of reproductive autonomy, so that it better reflects the new challenges arising from the increased use of genomic testing in pregnancy. We selected a qualitative interview approach for engagement with healthcare

professionals because this is an appropriate tool to explore the unknown terrain of their experiences and reflections in depth (Merriam and Tisdell 2016).

Participant recruitment

We sought to interview a range of healthcare professionals who care for pregnant women: general obstetricians, fetal medicine specialists, general practitioners, clinical geneticists, genetic counselors, and midwives. Participants were initially recruited through our professional networks, followed by snowball sampling (Noy 2008). Invitations to interview were sent via email. Purposeful sampling among the identified clinical specialties was also carried out through advertisements in professional newsletters and email lists, with potential participants emailing the research team to nominate their interest. Recruitment targeted healthcare professionals across Australia, including a mix of metropolitan, regional, and rural locations. It ceased once most locations, domains and specialties were represented and data saturation was achieved – recognizing the challenges of determining saturation in studies like this (Dunn *et al*. 2012). Research Ethics approval was granted by the University of Sydney Human Research Ethics Committee (2021/651). All participants provided written consent prior to interviews and were not remunerated.

Data collection and analysis

Semi-structured interview prompts (see Appendix 1) sought to uncover the following: the context in which prenatal testing is offered; the process by which results were received, interpreted and communicated; clinicians' knowledge of and attitudes towards the rapidly changing prenatal testing landscape (including how tensions between technological innovation and patient needs are negotiated); how reproductive autonomy is conceived of in the context of prenatal genomic testing, and barriers to and facilitators of its operationalization; and perspectives on the future of genomic testing and how to safeguard reproductive autonomy.

A total of 25 semi structured interviews (conducted by KD) were completed between May and December 2022. Almost all interviews were conducted via video conferencing software Zoom (one interview was conducted by phone), with interviews ranging between 42 min and one hour in length (median 50 min). Participants ranged in age from 25 to 59 years of age (median 43), and their professional experience ranged from one year to over 30 years (median 11), with all specialties in relation to prenatal care represented. Our sample included healthcare professionals based in states and territories outside of the (more populous) Eastern Australian states, metropolitan, regional or rural areas, and within public hospitals, private practice and IVF clinics (see Table 1).

Interviews were professionally transcribed, then verified and anonymized by LK. De-identified transcripts were managed in NVivo software (QSR

Table 1. Demographic characteristics of participants.

Characteristics	Total ($n = 25$)
Gender	
Man	7 (28%)
Woman	17 (68%)
Non-binary	1 (4%)
Age	
20–29 years	3 (12%)
30–39 years	6 (24%)
40–49 years	10 (40%)
50–59 years	6 (24%)
Professional Occupation	
Clinical geneticist	5 (20%)
Genetic counselor	5 (20%)
General practitioner	4 (16%)
Midwife	4 (16%)
Obstetrician (including Maternal Fetal Medicine Specialists)	7 (28%)
Years of practice	
1–5 years	7 (28%)
6–10 years	5 (20%)
10–20 years	7 (28%)
20 + years	6 (24%)
*Workplace**	
Public hospital	15
Private practice	11
State/territory of work	
Australian Capital Territory	2 (8%)
New South Wales	11 (44%)
Northern Territory	0 (0%)
Queensland	1 (4%)
South Australia	0 (0%)
Tasmania	2 (8%)
Victoria	6 (24%)
Western Australia	3 (12%)
*Location of work**	
Urban	22
Regional	3
Rural	2

*Categories are not mutually exclusive, so no percentages are provided

International). Data analysis followed a thematic analysis approach (Braun and Clarke 2022). The coding frame was developed with multidisciplinary input from the research team, who have backgrounds in bioethics, philosophy, genetic counseling, genomics, law, medicine, disability research, health services research, and qualitative research. Coding was undertaken by LK and KJL, and coding agreement and inter-coder reliability established by KD based on pilot coding.

Numerous provisional themes were identified through a process of open thematic mapping and repeated re-examination of the coded data, and refined provisional themes were then retested against the data.

In this article, we report data taken from coding that explores the context in which prenatal testing is offered, perceptions of its benefits and utility, and facilitators and barriers to the operationalization or realization of reproductive autonomy in a rapidly changing technological landscape.

Results

All participants discussed a range of factors and drivers that shape reproductive autonomy in the context of genomic testing in pregnancy. The themes identified capture the diversity of perspectives on the attraction of genomic information in pregnancy, how technology is rendering genomic information ubiquitous, and, crucially, the ways in which possibly overwhelming information provision may paradoxically constrain reproductive autonomy. Where illustrative quotes are used, respondents are identified by their professional occupation.

The dual attraction of genomic information in pregnancy: information and reassurance

Participants recognized both the clinical benefits of providing an actionable or informative "answer" through prenatal testing, and the value of being able to offer reassurance to pregnant women when no anomaly is identified:

> There's the clinical component to doing it, [namely to see] if you come up with something that might direct you to do further testing. Or, there's the social, emotional, mental health reasons to do it, [that is] for the woman who wants to hopefully be reassured that things are, you know, almost normal. (P3, Clinical Geneticist)

The presumed clinical benefits of information

A primary reason for offering genomic testing in pregnancy suggested by participants was to generate information relevant to fetal health. This enables healthcare professionals to serve the needs of pregnant women to have a "fuller understanding of the health of their baby and their pregnancy" (P12, Clinical Geneticist). It was also assumed that pregnant women would want to know any and all "problems":

> … there is a perception of things that are … routinely offered testing for in pregnancy, that those conditions are a problem, and those disabilities are significant. (P1, Genetic Counselor)

Genomic information was considered to be meaningful to decision-making, whether a decision to terminate an affected pregnancy or to prepare for a child with a genetic condition. In this sense, information is presumed to offer certainty

and to facilitate a course of action. Such action (making a choice) could be construed as evidence that autonomy was exercised:

> We offer it so that people have reproductive choice, so that they can choose what to do with the information[…]. (P16, Maternal Fetal Medicine Specialist)

Using genomic information to resolve uncertainty was also identified as an important driver of testing. This reflects the dominant view that "a lot of healthcare practitioners, they come from a point of view where their innate nature is that knowledge is power" (P2, Clinical Geneticist). Consequently, the value of newer tests, such as NIPT, is that they facilitate patient choice at the earliest possible point in pregnancy: "And I think there is a push to do that, you know, because otherwise there is this uncertainty [while awaiting further testing]" (P1, Genetic Counselor). For some practitioners, all information is valuable, and testing in pregnancy drives certainty. This belief may explain why healthcare professionals are increasingly offering prenatal testing to their patients, as one more critical participant observed:

> I honestly believe that the majority of those tests are driven by medical professionals. (P24, Genetic Counselor).

In addition, this drive is underpinned by a perceived responsibility of healthcare professionals to quantify probability to ensure that pregnant people are aware of their chances of having a child with a genetic or chromosomal condition: " … so it's our obligation as clinicians to say to women, 'Hey, there's this kind of testing that's available, you've heard of these genetic conditions'" (P13, Midwife). This suggests that the role of prenatal testing is to provide pregnant women any and all information regarding the probability that the pregnancy is not "normal":

> I take on the point of view that a lot of the obstetrics societies and things like that have these days, and that is: 'this is testing available to women to find out the genetic nature of their pregnancy', with the concept that we have framed these things as 'they're anomalies', they're, 'they're not normal'. (P13, Midwife)

> […] everybody's entitled to have the opportunity to get the piece of information if they want it. (P13, Midwife)

Accordingly, the practice of offering prenatal genomic testing becomes normalized for healthcare professionals, based on their expectation that pregnant people would prefer a "typical" child:

> So, partly I think it's a bit of a reflex. […] Partly it's because the whole general population has a much lower tolerance for an anomaly and for difference and for the extra burden that it creates (P19, Maternal Fetal Medicine Specialist).

The logic that sees genetic information as useful because it leads to actionable decision making, thereby increasing autonomy, also works in reverse. Some participants felt not all information was beneficial, in particular if it could not be used to facilitate action. For instance, one participant stated: "[it is not] okay to be offering all of this information if you then can't, sort of, do anything about it" (P12, Clinical Geneticist). Ultimately, the increased routinization of the offer of tests like NIPT was even seen to hinder a pregnant women's decision-making:

> I think sometimes it's not even their [pregnant women's] decision. I think sometimes, you know, some obstetricians … they have to have like a range of tests and then this is just kind of a routine test. I don't know if it's always like a decision that they [pregnant women] make or even if it is their decision, I'm not sure how informed their decision is. (P5, Genetic Counselor)

The allure of reassurance

While participants identified how prenatal genomic information can generate certainty, they also perceived that pregnant people seek tests like NIPT for reassurance.

> Well, I guess, number one, [they] probably want reassurance that their baby is healthy and maybe a sense of slightly more control over the process. (P20, General Practitioner)99% [of pregnant women], they just use it as a screening tool, in the same way as kind of an ultrasound. (P4, Clinical Geneticist).

It was also assumed that pregnant women are risk averse, and so undertake prenatal testing in aim to reduce the chance that they will have a child with a genetic or chromosomal condition:

> … humans hate risk. They hate the risk of having a not perfect baby and they hate the risk of basically invasive testing. So, $500 gets a maternal serum blood test and I can wipe away pretty much all of my risk … . (P13, Midwife)

But participants also noted that the increased volume and complexity of information that genomics can generate means pregnant people may receive results that are unanticipated and not reassuring:

> … a lot of people don't really think [about an issue being identified] … problems happen to other people […] They don't really think it's going to happen to them, but they'll just [take the test] anyway because that's what you do. And then when something is found, you know, then they're, yeah, they didn't really anticipate it. You kind of go for reassurance more than going looking for a problem. (P10, Obstetrician)

New tests can increase the risk of false reassurance. Participants described how they need to negotiate patients' perceptions that genomic information gleaned

from tests like NIPT was more accurate than information from other tests, such as ultrasound: "I think people think it's a better test" (P16, Obstetrician). Indeed, the limited coverage of NIPT can provide false reassurance:

> So, for example, if we find something really unusual in the morphology scan and we start talking about genetic testing, they tell, "Okay, I've already had genetic testing", which is the NIPT. "So, everything is normal in the baby." So, they are like falsely reassured in many situations. (P4, Clinical Geneticist)

The technological landscape of prenatal testing and the ubiquity of (more and more) information

Participants' responses revealed concerns over the pace of technology in providing genomic information in pregnancy, with more and more finely grained information becoming accessible. At the same time, the greater volume, complexity, and granularity afforded by genome-based tests can be so overwhelming as to negate any potential reassurance that can ever be provided by healthcare professionals.

The pace and drivers of change

Technologies that facilitate genomic testing were generally endorsed by participants. However, the extent to which more detailed (finer grained) information through expanded testing should be provided was less clear. One issue is the pace of technological development, and the attendant offer of unending growth in testing:

> It feels like it's moving really fast. We have been having lots of chats in our department recently about the speed with which the technology is evolving. The testing is becoming, you know, just 'wanted' by patients but also by their providers, like obstetricians mentioning the availability of more and more complex tests like prenatal exomes. (P1, Genetic Counselor)

Additional worries were voiced that the increasing availability of more expansive testing and more finely grained information could – paradoxically – increase uncertainty:

> So, if we look at case in point, let's look at microarray in pregnancy. I think it's an excellent test to detect significant chromosome abnormalities that *can* impact on prognosis, but if done as a routine screen, then the chances of picking up, you know, a variant of unknown significance is quite high relative to the chances of picking up a variant of known significance. (P3, Genetic Counselor)

At the same time, increasingly fine-grained test results make it "harder and harder to not offer people exomes for lots of things that we might not have before" (P1, Genetic Counselor). Accordingly, participants expressed concern that such tests

were being introduced without a sound rationale and in reaction to an uninterrogated demand:

> [This] is an example of where the technology became available and popular and by demand before the science really proved its use. So, you have a test that's sort-of there by popular demand, if you like, rather than because science evaluated it as being necessary. […]. (P3, Clinical Geneticist)

Consequences of testing amid limited resources for interpreting information

With more genomic technologies being adopted into prenatal care, participants felt that the increased provision of large amounts of complex genomic information demands significantly higher genomic literacy from both healthcare professionals and those having testing:

> So, all those nuances, like some [brands of NIPT] will have a tick box: 'I want this disease, I want this disease,' and even people who are highly educated with a medical background often are like: 'I don't know, I don't know anything about these conditions.' And so some of the other tests just say: 'we report on what we report on, you know, you get it or you don't.' It's a bit more black and white and I think all that variation within the field is pretty challenging for patients and people ordering it. (P12, Clinical Geneticist)

Participants felt that their patients' knowledge and understanding of genomic tests in pregnancy was often limited. Those choosing testing do not always have the capacity or resources to manage the nuances of this information, a problem amplified by the diversity of test offers and information available in the Australian test marketplace. Some pregnant people will not have the capacity to decide how useful testing will be to them without good support:

> I think there's just such a wide range of people from highly educated to no health literacy at all, and so it's just such a hodgepodge of different experiences […]. For some people are very aware and going into it with their eyes open and making their own informed choices and their own decisions, and then other people are operating on a very simple level … they just want reassurance that everything is okay and then are quite devastated when they go through a bit of a journey […] Or, they are not even at that level, and they will just go along with whatever their doctor says or their doctor may even just have a check list and it just gets done. (P25, Clinical Geneticist)

> So, I think they know a bit, but just maybe not enough. […] I think they just don't know the intricacies of the tests, like that it will pick up other things, that it might pick up things that they weren't expecting […] but not a genetic diagnosis in the baby. So, I think they have a basic understanding … that they're having it for abnormalities in the baby, and that they can choose to make pregnancy decisions around it, but maybe just not all that fine detail (P12, Clinical Geneticist).

> We test for 500 things versus this one only test for three things, so I do find that patients' understanding of how good these tests are, and what they are actually good for, is *super* variable, and women come to me with all sorts of different ideas about what NIPT *actually* can tell them. (P13, Midwife)

Without specialist pre-test counseling, the distress and anxiety of managing results from testing requires professional intervention *before* a decision can be made:

> When that test is ordered outside of a genetics expert, generally the requester has no idea what it means or how to explain it and we get couples referred who are extremely anxious and distressed, and we have to undo a lot of anxiety, when the truth is, you know, in the context of finding in a normal pregnancy with a normal anatomy scan and nothing else to suggest a problem, it's probably nothing, and to try and undo a lot of, you know, anxiety can be quite tricky. So, these tests definitely have a double edge to them. (Participant 3 –Clinical Geneticist)

This gap reflects that rapid technological development may be outpacing the resources required to facilitate reproductive autonomy. Participants were concerned about how healthcare professionals, and indeed the health system, manage this amount and detail of information. The current healthcare system in Australia is too resource-constrained to effectively manage the offering of (more and more) genetic information. There was a particular concern that the testing technology is running ahead of knowledge about how counseling should be provided. This places strain on health care professionals' capacity both to understand testing offers and communicate about them effectively:

> … it's advancing at such a rate already that it's probably *ahead* of what we can really do appropriate counseling for. So, probably the biggest challenge *I find* is that we really don't, the system at large or health systems overall, give us enough time for a consultation to explain what can and can't be found and whether you 'should or shouldn't' be doing this particular testing … So, that worries me. (P21, General Practitioner)

Reflecting the view discussed above that genomic information is useful only if it enables action, for many participants "management" of a pregnancy through testing carried a strong implication that the pregnancy would be terminated if a genetic or chromosomal anomaly was identified. The option to use testing to prepare for the birth of a child with a genetic or chromosomal condition was less valued, at least by some participants:

> The importance of NIPT is in families who may *do something* if we find some chromosomal abnormality. There are families where even if we see things like Trisomy 21 or Trisomy 18, they don't alter the way they manage the pregnancy. So, in those families, NIPT is just going to be a waste of time and money. (P4, Clinical Geneticist)

However, other participants thought that framing testing only in terms of termination was problematic:

> I think the thing that causes me concern is that often these tests are framed purely around termination and that a lot of doctor's sort of see this as a 'you find out if there is something wrong, if there is something wrong you terminate', as opposed to 'we are doing these tests to find out information that will guide us in the path going forward'. (Participant 10 - Obstetrician)

Decision-making in contexts of (overwhelming) information provision

Routinely using genomic information to identify potential conditions in a fetus is perceived to provide options for action. Yet when reflecting on the intersection between a desire for knowledge amid the inherent uncertainty of genomic information, participants were conscious that sometimes all that can be offered to patients is a possibility. Even if a fetal genotype can be ascertained, current genomic knowledge is often unable to provide certainty about the child's future phenotype or quality of life. As such, genomic information in pregnancy may not always be able to meaningfully influence decisions:

> I think the problem is, you're looking at the genetics of this baby. So, what you're looking at, even when I can tell you: 'this amniocentesis has come back and your baby is affected with this particular genetic problem', almost all of them sit on a spectrum. I can't tell you where this baby sits within that spectrum. (P13, Midwife)

The use of NIPT to detect sex chromosome anomalies is one domain in which the apparent certainty of genetic information may be obscuring the complexity and unpredictability of how that information will translate to lived experience:

> I think we really need some solid population data on what sex chromosome aneuploidies *are*. […] We need a description of the phenotype. (P19, Maternal Fetal Medicine Specialist)

Furthermore, if the appeal of testing for pregnant people is to get reassurance, receiving a result that an anomaly has been identified can be challenging, and can subsume or outweigh any indeterminacy about how the genotype will manifest.

The increasing availability of, and apparent demand for, genomic information works to reinforce assumptions about its predictive value while simultaneously obscuring the actual unpredictability of the phenotype and the life that a future child might lead. That is to say, the more weight that is placed on genotype, the more *significant* genomic information becomes *because* of its presumed value. This, in turn, can direct both patients and professionals to focus to extremes of phenotypes:

> [E]verybody pretty much counsels as though the baby's going to sit on the outermost difficult part of the spectrum and they're going to be the most affected, so I don't

> know how you get that nuance into conversations that aren't 4 h long and don't belabor the point, as every kid has a problem [of some kind]. […] It's super, super complex. (P13, Midwife)

The current limitations of genomic information and certain test types, such as NIPT, were also perceived as being under-appreciated by pregnant women:

> Look, I think it depends on what 'no shades of grey' they are looking for. So, if they are looking for an answer around the genetics of their baby, to the best of our ability then they have to have an invasive test … But I think a lot of people don't appreciate that a 'normal' NIPT [result] is still grey. (P11, Obstetrician)

It is difficult to manage the dual, and potentially contradictory, aspects of certainty and uncertainty in pregnancy. Participants expressed their conviction that pregnant women are striving towards complete avoidance of risk. This difficulty will only increase as more finely grained genomic information is provided by current and future prenatal testing technologies:

> What are we going to do? Because it [is] soon going to be full genome testing, guaranteed, none of this Trisomy 18, 21, 13, rubbish. It's going to be like full gamut, and our scientists are amazing, and the science is amazing, what the hell, how do we implement it? How do we hold so much more grey zone, right? Because there are huge percentages of the population walking around with microdeletions that live completely functional lives […] but if you can't prove that someone has the same microdeletion and you are going to try and guarantee that that baby will be normal - because everyone wants the perfect normal baby - we are going to go down a fucking wormhole of shit storm. (Participant 11-Obstetrician)

Discussion

We have reported findings from qualitative interviews with healthcare professionals who care for pregnant women. We sought to explore these professionals' views and experiences of reproductive autonomy in the setting of offering or following-up from prenatal testing in a rapidly changing technological landscape, one in which testing is becoming increasingly routinized and which is also moving towards a potential offer of fetal genome sequencing in all pregnancies. Our participants discussed why prenatal testing is offered and taken up, the impact of novel testing technologies, and how these developments will change decision-making.

Participants explained how they offer testing to inform the management of a pregnancy through acquiring information about relevant conditions. In so doing, they value the information that testing can generate as part of their need for certainty (enough information) that can guide action. They also noted that pregnant people may seek this information for a rather different reason: to be reassured that everything is "normal." If an unanticipated condition is identified,

professionals need to spend more time counseling, providing detailed information about the identified condition and working through the available options with the patient. Our data suggest that practitioners work from a background assumption that prospective parents will have a strong preference for a "normal" baby, an assumption that is probably influenced by the prevailing cultural ableism as well as a pragmatic awareness of the limited support and resources available to most families with a disabled child.

Technological innovation in prenatal testing is happening at a fast pace, with numerous drivers. Testing happens more often, and the results that are generated can be hard for pregnant women to understand, let alone for practitioners to explain. Participants are concerned that current resourcing in Australia's healthcare system may be inadequate to support the provision, interpretation and communication of prenatal genomic information. There are gaps in professionals' ability to engage in the process of sense-making that accompanies translating information into knowledge, yet knowledge is key to enabling pregnant women to make effective decisions. Without a clear idea of its meaning, genomic information is subject to significance creep (see below), taking on a salience that goes beyond its actual value.

Our findings continue a trajectory of professional concern over increasing information provision in prenatal testing. Over 30 years ago, Tymstra (1989) described how Dutch women sought this testing to be reassured, despite these tests being unable to offer certainty about fetal health. Williams, Alderson, and Farsides (2002) described how UK healthcare professionals saw the offer of prenatal testing as, paradoxically, both expanding and limiting pregnant women's choices. These professionals also felt the expansion of screening to be inevitable and inexorable, and out of their control – a sentiment still shared by our participants. Alongside the myriad advances in the intervening decades, new concerns have arisen. There have been reports of professional wariness about information overload (Agatisa *et al*. 2018; Perrot and Horn 2022), and a skepticism about the utility of acquiring increasing amounts of genetic information (Markens 2013). Our concerns about resourcing for appropriate counseling and models of care are echoed by others (Agatisa *et al*. 2018; Gammon *et al*. 2016; McKinn *et al*. 2022; Perrot and Horn 2022). Our findings suggest that these concerns are more relevant than ever in an era where the volume, detail, and availability of prenatal genomic information is rapidly increasing.

Inspired by R.L. Ackoff's (1989) articulation of the data-information-knowledge-wisdom hierarchy (see also (Rowley 2007)), we suggest that genomic testing, including in pregnancy, generates data (such as a fetal genomic DNA sequence). This is then filtered to generate information (taken to mean data brought together and contextualized so as to be communicable), such as whether two or three copies of a particular chromosome are present. Information is then interpreted and placed in a report, which is then communicated to the pregnant person.[1]

But having access to genomic information in pregnancy will only lead to meaningful decisions (including meaningfully contributing to autonomy) if the right kinds of information are offered in the right amount, at an appropriate level of complexity and with enough time for reflection so as to facilitate sense-making (Johnston, Farrell, and Parens 2017; Kringle-Baer 2017; Scully, Porz, and Rehmann-Sutter 2007). Providing these things can potentially enable the transformation of genomic information by the pregnant person into a kind of knowledge which can be usefully deployed. Yet, there is an underlying assumption that these processes (from data to information, to then be transformed to knowledge) just happen when genetic data is provided. This is not the case, and the conflation of genetic data with information, or of information with knowledge, can lead to confusion surrounding the meaning and significance of test results (Shkedi-Rafid, Horton, and Lucassen 2021).

"Significance Creep," coined by Manson (2006), describes the attachment of special significance to genetic information that can occur when people conflate "information contained within DNA" with "information that is *about* DNA" or "may be derived from knowledge of DNA (and thus be about phenotypic traits)." It is knowledge rather than information (Dive and Newson 2018), that can best enable reproductive autonomy of pregnant women.

The ongoing expansion of the scope of prenatal testing despite the concerns noted by our participants and elsewhere is, we contend, an indicator that genomic information has acquired a special, and potentially disproportionate, *salience*. This salience is adopted by both pregnant people and the health professionals who care for them. Information (including genomic information) is said to be salient when it stands out from its setting or environment. Previous research has suggested that when people perceive information as particularly salient it can interfere with the pursuit of otherwise settled goals (e.g. Bordalo, Gennaioli, and Shleifer 2022; Enke 2020; Kahneman 2011). Additionally, information from technological sources and which is delivered by healthcare professionals "stands out" to patients and appears to carry a distinct weight or salience in pregnancy decision-making (Knight and Miller 2021; McCoyd 2010; McLeod and Sherwin 2000).

Prenatal genomic information may be perceived as especially salient because it is provided in the context of the expectation of a "normal" pregnancy. The healthcare professionals in our study saw that pregnant women expect to be reassured by genetic testing; they do not expect a genetic or chromosomal condition to be identified, perhaps because the scope of testing is not fully appreciated. The expectation that this information will provide reassurance means that its value is often assumed. Combined with the normalization of prenatal testing through increasing test use, genomic information takes on a disproportionate significance compared to other sources of information about the pregnancy. This focus on promoting certainty, actionability and reassurance means that other relevant considerations can be overlooked, such as whether a genetic finding will manifest at all, which

phenotypes other than the "extremes" are possible, or how data on genotype can be related to the quality of life the future child will lead. The "significance creep" (Manson 2006) of genomic data, in which its value is assumed, leaves little space to interrogate either the kind of information it offers, or whether this information can be transmuted to knowledge at all. Genomic information – especially if indicating an anomaly – cannot be ignored or unlearned (Werner-Lin *et al*. 2017). Here it is also helpful to be reminded that pregnant people's perceptions of what information is useful are shaped by socio-relational contexts (Mills 2015) and normative expectations.

As noted, the presupposition that genomic information can necessarily be transformed to knowledge makes operationalization of reproductive autonomy more complicated (Lewit-Mendes *et al*. 2023; Markens 2013). As our findings show, care for pregnant people is not currently set up to facilitate decision-making based on test results that are uncertain or was unexpected. Combined with increasing routinization, increasing identification of fetal genomic information that introduces high levels of uncertainty and nuance – "shades of grey" – makes autonomous decision making more difficult in the absence of knowledge and understanding.

What is of concern, then, is how the potential for reproductive autonomy will be shaped by the salience given to genetic information and how this affects sense-making in the development of knowledge. This problem has also been recognized in other genetic testing domains, such as oncology (Johnson *et al*. 2005). Put another way, the overemphasis of genomic information necessitates progression down a particular decision-making pathway – regardless of whether that was the reason testing was sought or offered in the first place. Some healthcare professionals in our study, for example, did not see that continuing with a pregnancy following the identification of an anomaly was making a choice as they did not see this as an action. On this view, testing was pointless because the expected action – termination – was not taken.

If it is true that the salience of information can disrupt reproductive autonomy, and if salience itself is an attribute constructed by the offer of information in prenatal testing, then more attention must be paid to the impact of genomic information on the exercise of reproductive autonomy. Models of reproductive autonomy fit for the genomic age in prenatal testing must account for how the salience of genomic information is constructed and build in opportunities to reconcile it with embodied knowledge. While it is beyond our scope to sketch such an account of reproductive autonomy in detail, some of our interview findings point to relationality playing a key role, as well as opportunities for pregnant women to discuss and clarify their aims and values in prenatal testing, and critical reflection by both practitioners and patients on the role that genomic information should play in decision making in pregnancy.

We acknowledge our study is subject to limitations in that it only examined the views of healthcare professionals, who are an important but not the only

stakeholder group in prenatal testing.[2] In addition, different healthcare specialties will tend to bring different perspectives and hold different norms, as well as differing in their scope of practice and the point in pregnancy at which they see people. Against this, the strengths of our study include that it is among the first to ask health professionals specifically about the rapid uptake of genomic testing in pregnancy. Our interdisciplinary stance also brings a diverse range of substantive and methodological perspectives to bear on this complex issue. Additionally, health care professionals were invited to reflect on their patients' experiences, preferences, and values, which will then be further explored in a separate arm of the study.

Our findings reveal that the context in which prenatal testing is offered has significantly altered as the technology enabling such testing (and resulting information) has advanced in its sophistication and complexity. As our participants observed, the exercise of reproductive autonomy is recognized as requiring not only access to quality information, but also time, and an acknowledgement of how the subjective, relational and emotional status of the patient impacts their ability to absorb and use information. Further, there is a concomitant need for support beyond information provision as part of wider decision-making processes (Seavilleklein 2009). The significance creep and salience of genomic information, combined with an expectation of normal findings on testing, need to be borne in mind as genomic prenatal testing becomes more ubiquitous. The rapidly changing context of prenatal genomic testing serves to highlight what was always the case: that reproductive autonomy cannot be conflated with information provision or expanded prenatal choice, nor can it be identified solely through the actions it facilitates. Our research suggests the need to revisit how genomic tests in pregnancy should be provided so as to uphold reproductive autonomy. Reproductive autonomy in the genomic age should be manifested through a nuanced, relational, contextualized negotiation between the pregnant woman, healthcare professional, technological capabilities, and societal expectations.

Acknowledgements

Thank you to all the people who participated in this research. Thanks also to Associate Professor Rosalind McDougall for her input into our wider project and her helpful comments on an earlier draft of this paper; and to Oluwadara Odusi for her research assistance with revisions to this paper following peer review

Disclosure statement

No potential conflict of interest was reported by the author(s).

Funding

This work was supported by the Australian Research Council, under [grant number DP2020006].

Notes

1. Due to the scope of this paper, we are not able to discuss wisdom here in any detail. However, it is relevant to questions over what constitutes autonomous decision-making in pregnancy. This question is the subject of ongoing conceptual research within our group.
2. Views and attitudes of parents and prospective parents (including pregnant women) will be sought separately.

Data availability statement

The data that support the findings of this study are available on request from the corresponding author, AJN. The data are not publicly available due to the dataset containing information that could compromise the privacy of research participants.

Consent

All participants in this study provided written informed consent to participate.

References

Ackoff, R. L. 1989. "From Data to Wisdom." *Journal of Applied Systems Analysis* 16: 3–9.

ACOG & SMFM. 2020. "Screening for Fetal Chromosomal Abnormalities: ACOG Practice Bulletin Summary, Number 226." *Obstetrics & Gynecology* 136 (4): 859–867. https://doi.org/10.1097/AOG.0000000000004107.

Agatisa, Patricia K., Mary Beth Mercer, Marissa Coleridge, and Ruth M. Farrell. 2018. "Genetic Counselors' Perspectives About Cell-Free DNA: Experiences, Challenges, and Expectations for Obstetricians." *Journal of Genetic Counseling* 27 (6): 1374–1385. https://doi.org/10.1007/s10897-018-0268-y.

Baldus, Marion. 2023. "'Overestimated Technology - Underestimated Consequences' - Reflections on Risks, Ethical Conflicts, and Social Disparities in the Handling of Non-Invasive Prenatal Tests (NIPTs)." *Medicine, Health Care, and Philosophy* 26 (2): 271–282. https://doi.org/10.1007/s11019-023-10143-1.

Benn, Peter, and Audrey R. Chapman. 2016. "Ethical and Practical Challenges in Providing Noninvasive Prenatal Testing for Chromosome Abnormalities: An Update." *Current Opinion in Obstetrics & Gynecology* 28 (2): 119–124. https://doi.org/10.1097/GCO.0000000000000254.

Bernhardt, Barbara A., Danielle Soucier, Karen Hanson, Melissa S. Savage, Laird Jackson, and Ronald J. Wapner. 2013. "Women's Experiences Receiving Abnormal Prenatal Chromosomal Microarray Testing Results." *Genetics in Medicine* 15 (2): 139–145. https://doi.org/10.1038/gim.2012.113.

Best, Sunayna, Karen Wou, Neeta Vora, Ignatia B. Van der Veyver, Ronald Wapner, and Lyn S. Chitty. 2018. "Promises, Pitfalls and Practicalities of Prenatal Whole Exome Sequencing." *Prenatal Diagnosis* 38 (1): 10–19. https://doi.org/10.1002/pd.5102.

Bordalo, Pedro, Nicola Gennaioli, and Andrei Shleifer. 2022. "Salience." *Annual Review of Economics* 14 (1): 521–544. https://doi.org/10.1146/annurev-economics-051520-011616.

Bowman-Smart, Hilary, Julian Savulescu, Cara Mand, Christopher Gyngell, Mark D. Pertile, Sharon Lewis, and Martin B. Delatycki. 2019. "'Small Cost to Pay for Peace of Mind': Women's Experiences with Non-Invasive Prenatal Testing." *Australian and New Zealand Journal of Obstetrics and Gynaecology* 59 (5): 649–655. https://doi.org/10.1111/ajo.12945.

Braun, Virginia, and Victoria Clarke. 2022. *Thematic Analysis: A Practical Guide*. London: SAGE Publications Ltd.

Bull, Marilyn J. 2020. "Down Syndrome." *New England Journal of Medicine* 382 (24): 2344–2352. https://doi.org/10.1056/NEJMra1706537.

Christiaens, Lieve, Lyn S. Chitty, and Sylvie Langlois. 2021. "Current Controversies in Prenatal Diagnosis: Expanded NIPT That Includes Conditions Other than Trisomies 13, 18, and 21 Should Be Offered." *Prenatal Diagnosis* 41 (10): 1316–1323. https://doi.org/10.1002/pd.5943.

de Jong, Antina, Wybo J. Dondorp, Merryn V. E. Macville, Christine E. M. de Die-Smulders, Jan M. M. van Lith, and Guido M. W. R. de Wert. 2014. "Microarrays as a Diagnostic Tool in Prenatal Screening Strategies: Ethical Reflection." *Human Genetics* 133 (2): 163–172. https://doi.org/10.1007/s00439-013-1365-5.

de Melo-Martín, Inmaculada. 2006. "Genetic Testing: The Appropriate Means for a Desired Goal?" *Journal of Bioethical Inquiry* 3 (3): 167–177. https://doi.org/10.1007/s11673-006-9015-4.

Dive, Lisa, and Ainsley J. Newson. 2018. "Reconceptualizing Autonomy for Bioethics." *Kennedy Institute of Ethics Journal* 28 (2): 171–203. https://doi.org/10.1353/ken.2018.0013.

Donchin, Anne. 1996. "Feminist Critiques of New Fertility Technologies: Implications for Social Policy." *Journal of Medicine and Philosophy* 21 (5): 475–498. https://doi.org/10.1093/jmp/21.5.475.

Donchin, Anne. 2001. "Understanding Autonomy Relationally: Toward a Reconfiguration of Bioethical Principles." *The Journal of Medicine and Philosophy* 26 (4): 365–386. https://doi.org/10.1076/jmep.26.4.365.3012.

Dondorp, W. J., G. C. M. L. Page-Christiaens, and G. M. W. R. de Wert. 2016. "Genomic Futures of Prenatal Screening: Ethical Reflection." *Clinical Genetics* 89 (5): 531–538. https://doi.org/10.1111/cge.12640.

Donley, Greer, Sara Chandros Hull, and Benjamin E. Berkman. 2012. "Prenatal Whole Genome Sequencing: Just Because We Can, Should We?" *The Hastings Center Report* 42 (4): 28–40. https://doi.org/10.1002/hast.50.

Dunn, Michael, Mark Sheehan, Tony Hope, and Michael Parker. 2012. "Toward Methodological Innovation in Empirical Ethics Research." *Cambridge Quarterly of Healthcare Ethics: CQ: The International Journal of Healthcare Ethics Committees* 21 (4): 466–480. https://doi.org/10.1017/S0963180112000242.

Enke, Benjamin. 2020. "What You See Is All There Is." *The Quarterly Journal of Economics* 135 (3): 1363–1398. https://doi.org/10.1093/qje/qjaa012.

Farrell, Ruth M. 2013. "Symposium: Women and Prenatal Genetic Testing in the 21st Century." *Health Matrix (Cleveland, Ohio: 1991)* 23 (1): 1–13.

Gabriel, Jazmine. 2017. "Zooming Out: Solidarity in the Moral Imagination of Genetic Counseling." In *Reproductive Ethics*, edited by Lisa Campo-Engelstein and Paul Burcher, 7–25. Cham: Springer International Publishing. https://doi.org/10.1007/978-3-319-52630-0_2.

Gammon, B. L., S. A. Kraft, M. Michie, and M. Allyse. 2016. "'I Think We've Got Too Many Tests!': Prenatal Providers' Reflections on Ethical and Clinical Challenges in the Practice Integration of Cell-Free DNA Screening." *Ethics, Medicine and Public Health* 2 (3): 334–342. https://doi.org/10.1016/j.jemep.2016.07.006.

Hashiloni-Dolev, Yael, Tamar Nov-Klaiman, and Aviad Raz. 2019. "Pandora's Pregnancy: NIPT, CMA, and Genome Sequencing—A New Era for Prenatal Genetic Testing." *Prenatal Diagnosis* 39 (10): 859–865. https://doi.org/10.1002/pd.5495.

Hildt, Elisabeth. 2002. "Autonomy and Freedom of Choice in Prenatal Genetic Diagnosis." *Medicine, Health Care and Philosophy* 5 (1): 65–72. https://doi.org/10.1023/A:1014262809712.

Hodgson, Jan, and Merle Spriggs. 2005. "A Practical Account of Autonomy: Why Genetic Counseling Is Especially Well Suited to the Facilitation of Informed Autonomous Decision Making." *Journal of Genetic Counseling* 14 (2): 89–97. https://doi.org/10.1007/s10897-005-4067-x.

Holloway, Kelly, Nicole Simms, Robin Z. Hayeems, and Fiona A. Miller. 2022. "The Market in Non-invasive Prenatal Tests and the Message to Consumers: Exploring Responsibility." *The Hastings Center Report* 52 (2): 49–57. https://doi.org/10.1002/hast.1329.

Hui, Lisa, and Jane Halliday. 2023. "A Decade of Non-Invasive Prenatal Screening in Australia: National Impact on Prenatal Screening and Diagnostic Testing." *Australian and New Zealand Journal of Obstetrics and Gynaecology* 63 (2): 264–267. https://doi.org/10.1111/ajo.13638.

Hunt, Linda M., Katherine B. de Voogd, and Heide Castañeda. 2005. "The Routine and the Traumatic in Prenatal Genetic Diagnosis: Does Clinical Information Inform Patient Decision-Making?" *Patient Education and Counseling* 56 (3): 302–312. https://doi.org/10.1016/j.pec.2004.03.004.

Jackson, Emily. 2001. *Regulating Reproduction: Law, Technology and Autonomy.* London: Hart Publishing.

Johnson, J. David, Donald O. Case, James E. Andrews, and Suzanne L. Allard. 2005. "Genomics—the Perfect Information–Seeking Research Problem." *Journal of Health Communication* 10 (4): 323–329. https://doi.org/10.1080/10810730590950048.

Johnston, Josephine, Ruth Farrell, and Erik Parens. 2017. "Supporting Women's Autonomy in Prenatal Testing." *The New England Journal of Medicine* 377 (6): 505–507. https://doi.org/10.1056/NEJMp1703425.

Kahneman, D. 2011. *Thinking, Fast and Slow.* New York, NY: Macmillan.

Kaposy, Chris. 2017. "Noninvasive Prenatal Whole-Genome Sequencing: A Solution in Search of a Problem." *The American Journal of Bioethics* 17 (1): 42–44. https://doi.org/10.1080/15265161.2016.1251650.

Kaposy, Chris. 2023. "Prospects for Limiting Access to Prenatal Genetic Information about Down Syndrome in Light of the Expansion of Prenatal Genomics." *The New Bioethics* 29 (3): 226–246. https://doi.org/10.1080/20502877.2022.2130720.

Katz Rothman, B. 1986. *The Tentative Pregnancy: Prenatal Diagnosis and the Future of Motherhood.* New York: Viking Penguin.

Knight, Amber, and Joshua Miller. 2021. "Prenatal Genetic Screening, Epistemic Justice, and Reproductive Autonomy." *Hypatia* 36 (1): 1–21. https://doi.org/10.1017/hyp.2020.50.

Kringle-Baer, Kirsten M. 2017. "The Decision to Know: Pregnancy and Epistemic Harm." In *Reproductive Ethics*, edited by Lisa Campo-Engelstein and Paul Burcher, 45–60. Cham: Springer International Publishing. https://doi.org/10.1007/978-3-319-52630-0_4.

Lewit-Mendes, Miranda F., Hazel Robson, Joanne Kelley, Justine Elliott, Erica Brown, Melody Menezes, and Alison D. Archibald. 2023. "Experiences of Receiving an Increased Chance of Sex Chromosome Aneuploidy Result from Non-Invasive Prenatal Testing in Australia: 'A More Complicated Scenario than What I Had Ever Realized.'" *Journal of Genetic Counseling* 32 (1): 213–223. https://doi.org/10.1002/jgc4.1635.

Lippman, Abby. 1999. "Embodied Knowledge and Making Sense of Prenatal Diagnosis." *Journal of Genetic Counseling* 8 (5): 255–274. https://doi.org/10.1023/A:1022901131305.

Löwy, Ilana. 2017. *Imperfect Pregnancies: A History of Birth Defects and Prenatal Diagnosis*. Baltimore, MD: Johns Hopkins University Press.

Löwy, Ilana. 2022. "Non-Invasive Prenatal Testing: A Diagnostic Innovation Shaped by Commercial Interests and the Regulation Conundrum." *Social Science & Medicine* 304 (May): 113064. https://doi.org/10.1016/j.socscimed.2020.113064.

Manson, Neil C. 2006. "What Is Genetic Information, and Why Is It Significant? A Contextual, Contrastive, Approach." *Journal of Applied Philosophy* 23 (1): 1–16. https://doi.org/10.1111/j.1468-5930.2006.00317.x.

Markens, Susan. 2013. "'It Just Becomes Much More Complicated': Genetic Counselors' Views on Genetics and Prenatal Testing." *New Genetics and Society* 32 (3): 302–321. https://doi.org/10.1080/14636778.2013.822174.

McCoyd, Judith L. M. 2010. "Authoritative Knowledge, the Technological Imperative and Women's Responses to Prenatal Diagnostic Technologies." *Culture, Medicine, and Psychiatry* 34 (4): 590–614. https://doi.org/10.1007/s11013-010-9189-4.

McKinn, Shannon, Nasrin Javid, Ainsley J. Newson, Lucinda Freeman, Carissa Bonner, Antonia W. Shand, Natasha Nassar, and Katy J. L. Bell. 2022. "Clinician Views and Experiences of Non-Invasive Prenatal Genetic Screening Tests in Australia." *Australian and New Zealand Journal of Obstetrics and Gynaecology* 62 (6): 830–837. https://doi.org/10.1111/ajo.13533.

McLeod, Carolyn. 2002. *Self-Trust and Reproductive Autonomy*. Cambridge, MA: MIT Press.

McLeod, Carolyn, and Susan Sherwin. 2000. "Relational Autonomy, Self-Trust, and Health Care for Patients Who Are Oppressed." In *Relational Autonomy: Feminist Perspectives on Autonomy, Agency and the Social Self*, edited by Carolyn McLeod, and Natalie Stoljar, 259–279. Oxford: Oxford University Press.

Merriam, Sharan B., and Elizabeth J. Tisdell. 2016. *Qualitative Research: A Guide to Design and Implementation*. 4th ed. San Fransisco: Jossey Bass, Wiley.

Mills, Catherine. 2015. "Resisting Biopolitics, Resisting Freedom: Prenatal Testing and Choice." In *Resisting Biopolitics: Philosophical, Political, and Performative Strategies*, edited by S. E. Wilmer and Audronė Žukauskaitė, 121–134. London: Routledge.

Murdoch, Blake, Vardit Ravitsky, Ubaka Ogbogu, Sarah Ali-Khan, Gabrielle Bertier, Stanislav Birko, Tania Bubela, *et al*. 2017. "Non-Invasive Prenatal Testing and the Unveiling of an Impaired Translation Process." *Journal of Obstetrics and Gynaecology Canada* 39 (1): 10–17. https://doi.org/10.1016/j.jogc.2016.09.004.

Nature. 2019. "Early Detection: Prospective Parents Should Be Prepared for a Surge in Genetic Data (Editorial)" *Nature* 565 (7741): 536–536. https://doi.org/10.1038/d41586-019-00331-6.

Navon, Daniel, and Gareth Thomas. 2021. "Screening Before We Know: Radical Uncertainties in Expanded Prenatal Genetics." *OBM Genetics* 5 (4): 140. https://doi.org/10.21926/obm.genet.2104140.

Noy, Chaim. 2008. "Sampling Knowledge: The Hermeneutics of Snowball Sampling in Qualitative Research." *International Journal of Social Research Methodology* 11 (4): 327–344. https://doi.org/10.1080/13645570701401305.

Perrot, Adeline, and Ruth Horn. 2022. "Health Professionals and Scientists' Views on Genome-Wide NIPT in the French Public Health System: Critical Analysis of the Ethical Issues Raised by Prenatal Genomics." *PLoS One* 17 (11): e0277010. https://doi.org/10.1371/journal.pone.0277010.

Pös, Ondrej, Jaroslav Budiš, and Tomáš Szemes. 2019. "Recent Trends in Prenatal Genetic Screening and Testing." *F1000Research* 8: F1000 Faculty Rev-764. https://doi.org/10.12688/f1000research.16837.1.

Ravitsky, Vardit, Francois Rousseau, and Anne-Marie Laberge. 2017. "Providing Unrestricted Access to Prenatal Testing Does Not Translate to Enhanced Autonomy." *The American Journal of Bioethics* 17 (1): 39–41. https://doi.org/10.1080/15265161.2016.1251651.

Ravitsky, Vardit, Marie-Christine Roy, Hazar Haidar, Lidewij Henneman, John Marshall, Ainsley J. Newson, Olivia M.Y. Ngan, and Tamar Nov-Klaiman. 2021. "The Emergence and Global Spread of Noninvasive Prenatal Testing." *Annual Review of Genomics and Human Genetics* 22 (1): 309–338. https://doi.org/10.1146/annurev-genom-083118-015053.

Richardson, Anastasia, and Kelly E. Ormond. 2018. "Ethical Considerations in Prenatal Testing: Genomic Testing and Medical Uncertainty." *Seminars in Fetal and Neonatal Medicine* 23 (1): 1–6. https://doi.org/10.1016/j.siny.2017.10.001.

Rowley, Jennifer. 2007. "The Wisdom Hierarchy: Representations of the DIKW Hierarchy." *Journal of Information Science* 33 (2): 163–180. https://doi.org/10.1177/0165551506070706.

Schmitz, Dagmar. 2013. "A New Era in Prenatal Testing: Are We Prepared?" *Medicine, Health Care and Philosophy* 16 (3): 357–364. https://doi.org/10.1007/s11019-012-9411-y.

Schmitz, Dagmar, and Wolfram Henn. 2022. "The Fetus in the Age of the Genome." *Human Genetics* 141 (5): 1017–1026. https://doi.org/10.1007/s00439-021-02348-2.
Scully, Jackie Leach, Rouven Porz, and Christoph Rehmann-Sutter. 2007. "'You Don't Make Genetic Test Decisions from One Day to the next'–Using Time to Preserve Moral Space." *Bioethics* 21 (4): 208–217. https://doi.org/10.1111/j.1467-8519.2007.00546.x.
Seavilleklein, Victoria. 2009. "Challenging the Rhetoric of Choice in Prenatal Screening." *Bioethics* 23 (1): 68–77. https://doi.org/10.1111/j.1467-8519.2008.00674.x.
Shkedi-Rafid, Shiri, Rachel Horton, and Anneke Lucassen. 2021. "What Is the Meaning of a 'Genomic Result' in the Context of Pregnancy?" *European Journal of Human Genetics* 29 (2): 225–230. https://doi.org/10.1038/s41431-020-00722-8.
Suter, Sonia M. 2018. "The Tyranny of Choice: Reproductive Selection in the Future." *Journal of Law and the Biosciences* 5 (2): 262–300. https://doi.org/10.1093/jlb/lsy014.
Tymstra, T. 1989. "The Imperative Character of Medical Technology and the Meaning of 'Anticipated Decision Regret.'." *International Journal of Technology Assessment in Health Care* 5 (2): 207–213. https://doi.org/10.1017/s0266462300006437.
Van den Veyver, Ignatia B. 2016. "Recent Advances in Prenatal Genetic Screening and Testing." *F1000Research* 5: 2591. https://doi.org/10.12688/f1000research.9215.1.
Werner-Lin, Allison, Judith L. M. Mccoyd, and Barbara A. Bernhardt. 2019. "Actions and Uncertainty: How Prenatally Diagnosed Variants of Uncertain Significance Become Actionable." *The Hastings Center Report* 49 (Suppl 1 (May)): S61–S71. https://doi.org/10.1002/hast.1018.
Werner-Lin, Allison, Sarah Walser, Frances K. Barg, and Barbara A. Bernhardt. 2017. "'They Can't Find Anything Wrong with Him, Yet': Mothers' Experiences of Parenting an Infant with a Prenatally Diagnosed Copy Number Variant (CNV)." *American Journal of Medical Genetics Part A* 173 (2): 444–451. https://doi.org/10.1002/ajmg.a.38042.
Williams, Clare, Priscilla Alderson, and Bobbie Farsides. 2002. "Too Many Choices? Hospital and Community Staff Reflect on the Future of Prenatal Screening." *Social Science & Medicine* 55 (5): 743–753. https://doi.org/10.1016/S0277-9536(01)00200-3.
Wise, Jacqui. 2022. "Non-Invasive Prenatal Screening Tests May Give False Results, Warns US Regulator." *BMJ* 377: o1031. https://doi.org/10.1136/bmj.o1031.

Appendix 1

Interview schedule
General

(1) Tell me a bit about yourself and your work

Prenatal testing experience

(2) In your experience, what is prenatal testing for? Why do you think it is offered?
(3) Are there any tests that you routinely offer? (Why?) Are there other tests that you wait for patients to mention? (Why?)

(4) What is your role in terms of patient interaction, talk me through when you would see pregnant people within the contexts of these tests?

Prenatal testing knowledge

(5) What's your take on how things are developing in prenatal testing at the moment? Has anything surprised you in relation to how the field is developing? Does anything worry you?
(6) Why do you think that pregnant people seek these tests? What is appealing about them?
(7) How well do you think pregnant people understand the kinds of testing available to them; what factors in the choices that they make; how do you think pregnant people make these choices?
(8) If you have concerns or worries in the course of your work, what do you generally do?

Reproductive Autonomy

The term reproductive autonomy is very commonly used now a days in this field. We are really interested in the wider implication of autonomy in reproductive genomics'.

(9) How do you understand reproductive autonomy? How, in the context of your work, would you go about defining or describing 'reproductive autonomy'?

What, in your view, should reproductive autonomy aim to achieve?

Could you give me an example of a case where you feel that the person or couple involved did exercise reproductive autonomy? Why was that?

Could you give me an example of a case where you feel that the person or couple involved did not exercise reproductive autonomy? Why was that?

(10) Do you think that, in general, pregnant people are making autonomous decisions about prenatal testing?

Are they in a good position to choose? Why do you think this is?

What kind of support do you think you should be providing to the people you care for? What are their needs? (informational, emotional, values clarification other?)

Do you think your description and views would align with those of your colleagues? How is this understood in your field?

(11) [If their answers are in line with the traditional literature] Your answers are very much in line with what we find in the traditional literature, which emphasizes individual decision-making …
(12) [If their answers are out of step with traditional literature] Your answers diverge in some interesting ways from what we find in the traditional literature, which emphasizes individual decision-making …

(13) In our project, we're developing ideas about reproductive autonomy that include recognizing the relationships we have with each other and the value of being able to consult with a range of people prior to making a decision.

How do you feel about this in respect to your work?
How would you go about this?
How feasible do you think this might be to use in practice?

The future

(14) Thinking about the future of prenatal testing, do you think anything needs to change?
(15) How do these changes intersect with how you think about reproductive autonomy?

Is there anything else you would like to raise that we have not mentioned already?