Unconditional access to non-invasive prenatal testing (NIPT) for adult-onset conditions: a defence

India R Marks ,[1] Catherine Mills,[2] Katrien Devolder [3]

[1]Faculty of Medicine, Nursing and Health Sciences, Monash University, Clayton, Victoria, Australia
[2]Monash Bioethics Centre, Monash University, Clayton, Victoria, Australia
[3]The Oxford Uehiro Centre for Practical Ethics, University of Oxford, Oxford, UK

Correspondence to
Dr India R Marks, Faculty of Medicine, Nursing and Health Sciences, Monash University, Clayton, VIC 3800, Australia; india.marks98@gmail.com

Received 3 March 2023
Accepted 18 April 2023
Published Online First 3 May 2023

Check for updates

© Author(s) (or their employer(s)) 2024. No commercial re-use. See rights and permissions. Published by BMJ.

To cite: Marks IR, Mills C, Devolder K. *J Med Ethics* 2024;**50**:102–107.

ABSTRACT

Over the past decade, non-invasive prenatal testing (NIPT) has been adopted into routine obstetric care to screen for fetal sex, trisomies 21, 18 and 13, sex chromosome aneuploidies and fetal sex determination. It is predicted that the scope of NIPT will be expanded in the future, including screening for adult-onset conditions (AOCs). Some ethicists have proposed that using NIPT to detect severe autosomal AOCs that cannot be prevented or treated, such as Huntington's disease, should only be offered to prospective parents who intend to terminate a pregnancy in the case of a positive result. We refer to this as the 'conditional access model' (CAM) for NIPT. We argue against CAM for NIPT to screen for Huntington's disease or any other AOC. Next, we present results from a study we conducted in Australia that explored NIPT users' attitudes regarding CAM in the context of NIPT for AOCs. We found that, despite overall support for NIPT for AOCs, most participants were not in favour of CAM for both preventable and non-preventable AOCs. Our findings are discussed in relation to our initial theoretical ethical theory and with other comparable empirical studies. We conclude that an 'unconditional access model' (UAM), which provides unrestricted access to NIPT for AOCs, is a morally preferable alternative that avoids both CAM's fundamental practical limitations and the limitations it places on parents' reproductive autonomy.

INTRODUCTION

Over the past decade, non-invasive prenatal testing (NIPT) has been widely adopted into routine obstetric care. NIPT analyses fetal cell-free DNA circulating in the maternal plasma and can therefore detect fetal genetic abnormalities.[1] The standard NIPT currently screens for trisomies 21, 18 and 13, sex chromosome aneuploidies and fetal sex determination. It is predicted, however, that the future scope of NIPT could be expanded to include screening for adult-onset conditions (AOCs), including, for example, early-onset Alzheimer's disease and hereditary breast cancer.[i]

One key ethical question that arises when contemplating using NIPT to screen for AOCs is whether it would be permissible to continue a pregnancy in the case of a positive test result.[ii] This has been considered particularly problematic in the case of severe, untreatable AOCs, such as Huntington's disease,[iii] not only because of the direct health impacts for the future child, but also because of potential harm they may experience due to the genetic knowledge obtained.

Because of these concerns, some ethicists have proposed to offer only 'conditional access' to prenatal genetic testing for Huntington's,[2–5] that is, to either strongly discourage, or simply not offer, prenatal testing for the disease if the prospective parents do not intend to terminate the pregnancy in the case of a positive result. For example, MacLeod *et al* have proposed that any 'couple requesting prenatal testing [for AOCs] must be clearly informed that if they intend to complete the pregnancy…there is no valid reason for performing the test'[3]. Likewise, in a debate by Duncan *et al*, Delatycki argues that 'if it is clear that a couple will not terminate…then the test should not be offered'.[6] We refer to such an approach to prenatal screening as the 'conditional access model' (CAM).

While one option would be to adopt CAM for NIPT for severe and untreatable AOCs, like Huntington's, there have also been calls to adopt CAM for a wider range of AOCs. Indeed, some guidelines, such as the 2016 position statement of the National Society of Genetic Counsellors, have recommended CAM for NIPT screening for any AOC.[7]

In this paper, we investigate these options. We argue that unrestricted access to NIPT for AOCs is preferable to CAM, and, presenting results from a recent survey, demonstrate that this position is likely to be consistent with community views. After first explaining CAM and its rationale, we outline its key ethical and practical limitations. We then explain why we think that the unrestricted access model (UAM) is preferable. We present study findings investigating the moral intuitions of NIPT users on the topic. Finally, we compare our empirical data with our ethical analysis, defend our conclusion and provide ideas for future direction of research.

THE CONDITIONAL ACCESS MODEL

'Conditional access' to testing has been proposed as a means of addressing the ethical predicament which arises when parents choose to continue a pregnancy prenatally diagnosed with Huntington's disease.[2–5] Based on CAM, parents would be informed that

[i]For the purposes of this paper, AOCs are defined as conditions which most commonly manifest over the age of 18 years.

[ii]This study does not discuss the ethics of termination in general, but instead assumes termination of pregnancy is legal and morally permissible in at least some circumstances.

[iii]Huntington's disease is a progressive brain disorder that results in problems with mental health, behaviour, movement and communication. Symptoms of Huntington's disease usually develop between ages 30 and 50. The disease gets gradually worse over time and is usually fatal after a period of up to 20 years.

J Med Ethics: first published as 10.1136/jme-2023-109070 on 3 May 2023. Downloaded from http://jme.bmj.com/ on November 30, 2025 at Zanjan University of Medical Sciences. Protected by copyright, including for uses related to text and data mining, AI training, and similar technologies.

there is no valid reason for performing the test if they intend to proceed with the pregnancy regardless of the result.[3]

CAM aims to protect the future child's autonomy by theoretically preventing the birth of a child who has its genetic status already known.[8] Specifically, it is motivated by concerns that the obtained genetic knowledge could result in the violation of the future child's 'right not to know', in adverse psychological sequelae for the future child and/or their parents, and in genetic discrimination against the future child.

The right not to know

The argument for CAM based on the future child's 'right not to know' their genetic information is predicated on the basis that some individuals may prefer ignorance to avoid any emotional burden associated with genetic knowledge.[9 10] Research shows that most adults who are at risk for Huntington's disease do not opt to undergo predictive testing.[11] Therefore, it is deduced that many at-risk children would not have autonomously chosen to undergo predictive testing if the decision had been left for them to make as an adult.

Psychosocial harm to the child

CAM has also been proposed as a way of preventing psychosocial harm resulting from the knowledge of one's genetic predisposition to an AOC. This position proposes that an awareness of a child's Huntington's disease status could result in psychosocial harms or impaired familial relationships: the child or parent(s) might become anxious or distressed knowing that the child is at risk of developing an AOC.[12 13] This could manifest in poor self-esteem in childhood, possible harm to the parent–child relationship or the 'vulnerable child phenomenon', whereby parents treat their child with overprotectiveness and extreme concern due to the child being 'at risk'.[7 14 15] Introducing CAM for NIPT for Huntington's disease would theoretically prevent this predicament.

Genetic discrimination

Other related concerns which would be avoided by CAM regard the possibility that individuals with a known predisposition to an AOC may be denied employment or insurance opportunities.[7 14 16 17] Personal knowledge of a genetic predisposition to an AOC would likely oblige the future person to inform employers or insurers, who could use this information in a discriminatory manner.

LIMITATIONS OF THE CAM

Having described the motivations behind CAM, we will next outline some of the limitations of this model. We argue these practical and ethical limitations deem CAM to be not only ethically problematic, but also ineffective and unsuited to clinical practice.

It is important to clarify that, in order to outline the limitations we see with CAM, we uphold that Huntington's disease presents a very strong case for CAM, as it is a lethal disease with no known treatment or cure. This gives us reason to believe that supposed harms such as psychosocial impacts, genetic discrimination and breach of the future child's rights can be reasonably presumed to manifest most strongly in the case of Huntington's disease screening. Based on this assumption, we consider that if CAM is unsuitable for Huntington's disease, there is good reason to believe that it is not suitable for milder or preventable AOCs either.

Unconvincing motivations for conditional access

While we acknowledge the validity of concerns around the future child's well-being were NIPT for AOCs to be permitted, importantly, there is a lack of robust evidence which exists to support these outcomes. Existing systematic reviews have found there is poor evidence to suggest that receiving predictive genetic screening information actually confers decreased psychosocial well-being in children.[18 19] There have even been suggestions that children could, in fact, experience psychosocial benefit, including reduced anxiety, reduced uncertainty about the future and ability to make more realistic life choices.[13 20 21] The lack of evidence here does not conclusively discredit the risk of psychosocial harm, and indeed, evidence may emerge in the future. Despite this, in the absence of strong evidence, the concern that the child would suffer psychologically from having knowledge about their AOC cannot currently provide a good reason to implement CAM.

Concerns about genetic discrimination are, too, speculative, and there are suggestions that they could be effectively addressed through structural reforms. For instance, Taylor-Sands and Bowman-Smart suggest that these concerns could be ameliorated through an independent review and amendment of current insurance and employment laws.[22] The US Genetic Information Non-Discrimination Act (GINA) of 2008 is an example of a legislative intervention that has arguably been reasonably successful in this regard.[23] This law prohibits health insurers and employers from discriminating based on genetic predisposition to a disease.[iv] Interestingly, data show that there have been very few claims filed to the relevant equal employment committee since GINA's introduction, and most claims that were made were dismissed due to lack of reasonable cause.[23] Therefore, GINA provides a case study of legislation which signals that fears of discrimination can be mitigated without placing restrictions on prospective parents who may wish to undertake NIPT for AOCs.

Adverse effects on the doctor–patient relationship

Another likely limitation of CAM is that it could motivate prospective parents to falsely promise to terminate a pregnancy simply to be granted access to the screening opportunity.[6] Undoubtedly, most parents would not want to mislead their healthcare provider, but CAM may compel them to behave (arguably) immorally by doing so and this may result in feelings of guilt. What's more, this scenario may erode existing patient-clinician trust, adversely impacting, and possibly permeating into other aspects of, the therapeutic relationship. In addition, in this scenario, CAM would often be ineffective at achieving its objective altogether, as parents who lied about their termination intentions would, in line with their true intentions, probably not terminate once genetic results were available to them.

Inability to predict termination intentions

Another key limitation arises from the difficulty for parents to accurately predict their future actions before knowing test results. Current evidence suggests that it is challenging to make an informed decision about termination before being presented with the reality of a diagnosis.[24] For instance, the proportion of women who indicate that they might consider termination for

[iv]Notably, in Australia no individual risk assessment occurs in the private health sector, meaning that genetic discrimination does not take place in the health insurance industry.[26] Therefore GINA, which specifically applies to health insurance, is not directly generalisable to an Australian context. Regardless, the potential for discrimination still exists in other Australian insurance sectors, such as life insurance and income protection insurance.[26]

J Med Ethics: first published as 10.1136/jme-2023-109070 on 3 May 2023. Downloaded from http://jme.bmj.com/ on November 30, 2025 at Zanjan University of Medical Sciences. Protected by copyright, including for uses related to text and data mining, AI training, and similar technologies.

Down's syndrome is far lower than the actual rate of termination among cases of confirmed Down's syndrome in pregnancy.[24] This suggests that using hypothetical termination intentions to guide conditional access to NIPT for AOCs would be an unreliable means of predicting the actual likelihood of termination. This stance has been reflected by the National Society of Genetic Counsellors: 'Answering hypothetical questions is not a genuine proxy for being confronted with real results…Expecting fully formed decisions will be made in advance is not a reasonable expectation'.[7]

Insufficient reason to restrict reproductive autonomy

Allowing parents to screen their fetus prenatally is a practice often based on the moral framework of reproductive autonomy.[25] If reproductive autonomy is what matters most, then, in the absence of any countervailing morally weighty considerations, it should be up to parents whether to use prenatal screening, regardless of their termination intentions.[4] We acknowledge, however, that reproductive autonomy could be constrained by other considerations if they were compelling enough. This could be based on a concept such as John Stuart Mill's 'Harm Principle', which holds that individuals should be free to act as they wish, unless these actions cause perceivable harm to others, which would justify placing restrictions on individual liberties.[26] Therefore, if continuing pregnancies known to be predisposed to AOCs was shown to result in significant harms, this could justify restricting reproductive autonomy through CAM.

In this case, however, we have shown that there are no such countervailing considerations, as concerns over harm to the future child seem to be largely unjustified according to the current evidence base. This, combined with the inherent practical limitations which would likely deem CAM ineffective anyway, leads us to conclude that CAM places an unjustified restriction on the reproductive autonomy of prospective parents.

Further, restricting parents' reproductive autonomy based on the future child's right not to know may also be unnecessary. In particular, we think implementing this model to protect this interest would fail to sufficiently consider other highly relevant interests. As we have outlined in another study, there are several benefits that parents and the family unit could derive from prenatal testing for AOCs, even without intent to terminate.[27] The child's interest in not knowing genetic information is one, but not automatically the only or dominating, consideration when making these prenatal decisions. The notion of justifiable intrusion on patient privacy is already evident in the medical realm; for example, under ethical guidance, patient confidentiality should broadly be upheld unless the clinician holds concerns for the safety of the patient or others. Thus, there are certain serious scenarios in which a nuanced consideration of competing interests allows another individual to rightfully violate a patient's privacy. We uphold that prospective parents should be given the opportunity to weigh their own interests and the interests of their future child (including a possible interest in not knowing) in order to make a decision about NIPT for AOCs. Thus, we conclude that to implement CAM and thereby restrict parents' reproductive autonomy would be largely unjustified.

The question then arises what alternative model for the use of NIPT for AOCs we should adopt. One alternative is unconditional access to NIPT for AOCs—a model that allows for open access to screening, regardless of one's intention to terminate. We will defend this model in the next section.

THE UNCONDITIONAL ACCESS MODEL

The UAM would involve offering the screening opportunity to all prospective parents regardless of termination intentions. This model would avoid the aforementioned limitations associated with CAM. Further, it would entail a parent-led decision-making process which acknowledges the reality that most medical decisions involve weighing up several interests and involve some trade-offs between these interests.[28] In this way, parents would attend to their own interests when making screening decisions, as well as acting as custodians for the future child's possible interests. Allowing parents to carry out this role would require heavy emphasis on high-quality, neutral pre-test counselling, allowing parents to consider the possible harms and benefits of undergoing screening and make informed judgements about the possible repercussions of choosing to reveal fetal information about AOCs.

In summary, we have highlighted several ethical and practical limitations of CAM, which significantly restrict its aims, and have instead advocated for UAM. The ideas from this philosophical discourse will be integrated with the empirical component of this paper, for which we collected data on the relevant views of NIPT users.

EMPIRICAL STUDY

Individuals who have used NIPT are direct stakeholders with unique views on the experiences of pregnancy and parenthood, and can add valuable insights around whether CAM would be morally acceptable in clinical practice. Moreover, integrating the views and experiences of people who are personally acquainted with this area of ethical practice ensures that the discourse is not prejudiced or severely limited by philosophical perspectives only. Finally, it could also assist in ensuring that relevant ethical policies are broadly aligned with prevailing public attitudes, a phenomenon known as 'moral pragmatics'.[29]

METHODS

Study design

A cross-sectional study design was used to survey stakeholder perspectives on NIPT.

Recruitment

Participants were individuals or partners of individuals who had previously undergone NIPT. They were recruited through advertisements posted on online forums and websites which distribute information and resources about fertility, pregnancy and parenting, namely: BubHub, EveryBump, BabyCenter, Melbourne Mums Groups and North Sydney Mums Group. Participants under 18 years and those who had never used NIPT were excluded.

Before providing consent, participants were directed to an explanatory statement containing information on what the study involved, inclusion and exclusion criteria, possible risks and benefits of participating, participant anonymity and data management and storage.

Survey

A study-specific survey was developed with assistance from a research team with expertise in bioethics and empirical research methodology. The survey was administered via Qualtrics, an online survey platform. Responses were collected between July and September 2021.

J Med Ethics: first published as 10.1136/jme-2023-109070 on 3 May 2023. Downloaded from http://jme.bmj.com/ on November 30, 2025 at Zanjan University of Medical Sciences. Protected by copyright, including for uses related to text and data mining, AI training, and similar technologies.

Survey data collected included information about participants' past experiences with NIPT, plus 40 items pertaining to participants' support for the availability of NIPT for particular traits/conditions (including preventable[v] and non-preventable[vi] AOCs), personal interest in testing for that trait/condition and personal likelihood of terminating a pregnancy based on that trait/condition. Participants were also asked to indicate their level of agreement with relevant ethical issues, including their support for CAM and their views on the ethical acceptability of continuing pregnancies known to be predisposed to AOCs. Item responses were measured on a 5-point Likert scale (definitely not, probably not, unsure, probably and definitely) to indicate level of agreement. Sociodemographic data were collected, which included age, gender, education level, household income, marital status and childbearing status.

Analysis

SPSS V.27 was used to analyse survey data. For analyses, 5-point Likert scales were collapsed to 3-point scales (positive, unsure and negative). Data were summarised using descriptive statistics (frequencies and percentages). The actual number of responses was used as the denominator (actual n) for the calculation of frequencies where there were missing responses. χ^2 tests were conducted to explore associations between demographic characteristics and support for NIPT screening for AOCs. A $p<0.05$ was considered statistically significant.

RESULTS

A total of 118 participants were recruited between July and September 2021. Nine were excluded due to significantly incomplete surveys. The final sample included 109 participants. Demographic characteristics are shown in table 1. Some of the collected survey data are reported in this study, with other survey results reported elsewhere in another related study.[27]

Table 1 Demographic characteristics of study cohort.

Participant characteristic		Participants (n)	Percentage
Age (years) n=109	18–25	2	1.8
	26–30	15	13.8
	31–35	50	45.9
	36–40	29	26.6
	41+	13	11.9
Gender n=96	Male	1	1.0
	Female	95	99.0
	Non-binary or other	0	0.0
Highest completed level of education n=96	Secondary school (year 10 or below)	1	1.0
	Secondary school (Victorian Certificate of Education or equivalent)	1	1.0
	Technical or trade certificate	10	10.4
	Bachelor's degree	40	41.7
	Postgraduate qualification (eg, Masters, PhD)	44	45.8
Combined household income after tax n=96	Less than US$25 000	0	0.0
	US$25 000–US$49 999	0	0.0
	US$50 000–US$99 999	9	9.4
	US$100 000–US$149 999	19	19.8
	US$150 000–US$199 999	25	26.0
	US$200 000–US$299 999	27	28.1
	More than US$300 000	12	12.5
	Prefer not to say	4	4.2
Marital status n=96	Single	1	1.0
	Partnered	18	18.8
	Married	77	80.2
Current no of children n=96	0	9	9.4
	1	51	53.1
	2	30	31.3
	3	5	5.2
	4	1	1.0
Pregnancy status n=96	Not pregnant	67	69.8
	Pregnant	27	28.1
	Unsure	2	2.1
Intends on having more children n=96	No	31	32.3
	Yes	45	46.9
	Unsure	20	20.8
Personal history of any conditions mentioned in the survey n=96	No	92	95.8
	Yes	4	4.2
Family history of any conditions mentioned in the survey n=96	No	82	85.4
	Yes	14	14.6

n=109 for age and n=96 for all other participant characteristics due to some incomplete survey responses, resulting in omitted demographic characteristics.

Views on NIPT for AOCs

Participants expressed strong support for novel NIPT screening for AOCs. Overall, 70.9% thought that NIPT should be available to find out about preventable AOCs, such as increased risk of bowel cancer. Similarly, 80.8% of respondents thought that NIPT should be available for non-preventable AOCs, such as Huntington's disease. No significant correlations were found between any demographic characteristics and support for NIPT for AOCs.

Views on the CAM

Most participants did not approve of CAM for both preventable and non-preventable AOCs, as displayed in figure 1.

Views on continuation of pregnancies diagnosed with AOCs

Most participants generally believed that it is ethically acceptable to continue a pregnancy where the fetus has been diagnosed with an AOC. Specifically, 90.6% of participants found it acceptable to continue pregnancies predisposed to preventable AOCs, with 4.2% finding this unacceptable and 5.2% unsure. Alternatively, 62.5% found it acceptable to continue pregnancies predisposed to non-preventable AOCs, with 13.5% finding this unacceptable and 24.0% unsure.[vii]

DISCUSSION

Based on our survey findings, most participants showed support for NIPT for both preventable and non-preventable AOCs. The vast majority of participants also did not support the implementation of CAM, aligning with our stance that this model is unsuitable for use in clinical practice.

[v]A preventable AOC is an AOC whereby the phenotype can be prevented from occurring through surveillance or therapeutic measures. The example given to survey participants was hereditary bowel cancer, which could be prevented through regular bowel cancer screening.

[vi]A non-preventable AOC is an AOC whereby the phenotype cannot be prevented from occurring. The example given to survey participants was Huntington's disease, for which there are no available surveillance or therapeutic measures that can be effectively used to avoid disease onset.

[vii]Data presented in this paper represents a subsection of total data collected: additional data from this survey is presented in another paper.

J Med Ethics: first published as 10.1136/jme-2023-109070 on 3 May 2023. Downloaded from http://jme.bmj.com/ on November 30, 2025 at Zanjan University of Medical Sciences. Protected by copyright, including for uses related to text and data mining, AI training, and similar technologies.

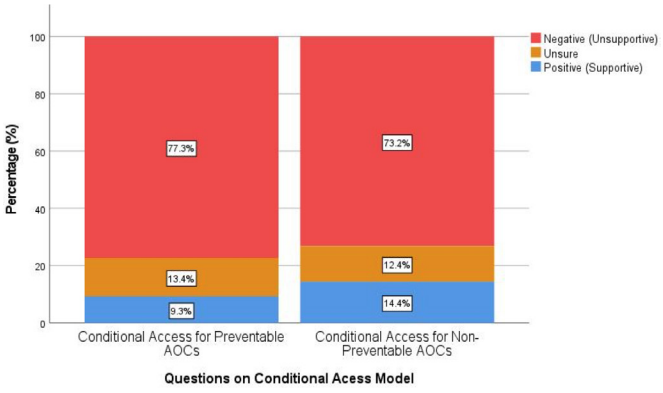

Figure 1 Participant support for the conditional access model for NIPT for adult-onset conditions (AOCs).
Figure displays frequencies (percentages) of the sample with positive, unsure and negative responses to questions asking about whether NIPT for preventable and non-preventable AOCs should only be offered to parent(s) who intend to terminate an affected pregnancy (n=97 for both questions).

Our survey findings related to support for NIPT for both preventable and non-preventable AOCs differ from other studies, which have generally shown lower parental support for NIPT for AOCs.[30–33] This finding might suggest that this sample places significant value in reproductive autonomy and recognises the benefits that this testing could yield for parents and the family unit.

The overall rejection of conditional access in our survey suggests that, from a policy perspective, this model would lack public support and therefore may not be feasible in practice. This result is consistent with, and could indeed be explained by, the sample's overall moral acceptance of the continuation of pregnancies diagnosed with AOCs. This finding likely indicates that many participants see no convincing need for conditional access: if continuing these pregnancies is deemed to be ethically acceptable, then restricting screening access only to parents who would terminate for an AOC is likely to be viewed as a superfluous measure.

Moreover, because most participants indicated that it is permissible to continue pregnancies known to be predisposed to AOCs, it can be inferred that these participants do not estimate that the possible harms of this scenario would outweigh the benefits. That is, most participants consider possible harms such as adverse psychosocial impacts on the future child and genetic discrimination to be minimal enough that they do not supersede the benefits that parents may experience from being able to practice reproductive autonomy. As we have shown in a related study, however, most participants still agreed that most future children and parents who are aware of their predisposition to AOCs would experience anxiety or distress as a result: most acknowledge a likelihood of some harm.[27] Thus, within the finding that continuing these pregnancies is acceptable among most participants is an implicit appraisal that either these harms are significantly outweighed by benefits, or simply should not be regulated by governing bodies. This finding ultimately champions reproductive autonomy in a similar manner as our theoretical discussion which advocates for UAM: despite acknowledging the possibility of harm, participants still believe that prospective parents should have the opportunity to undertake this screening based on their own implicit beliefs, values and appraisal of the situation.

The limitations of the empirical study need to be considered. First, participants were predominantly female and of high socioeconomic status. These factors reduce the generalisability of the findings. Bias may have also been introduced by the self-selection recruitment strategy and use of an online survey platform.

Future research could focus on collecting NIPT users' ethical intuitions on this topic with a larger, more diversified sample population. This could include surveying the broader public, not just NIPT users, which is relevant considering possible population impacts if this testing were to become widespread. Our survey did not question participants on exactly why they objected to CAM including specific limitations, which would be useful information for future research. In addition, as outlined earlier, evidence is lacking on the psychosocial experiences of children who are made aware of their genetic information about AOCs before they can autonomously make this decision for themselves. We suggest further research into this area to determine whether these harms are real and significant, and therefore, whether there is a convincing need to restrict the continuation of pregnancies known to be predisposed to AOCs.

CONCLUSION

In this study, we have concluded that conditional access is not an acceptable means of offering NIPT for AOCs to prospective parents. To reach this conclusion, we integrated both theoretical and empirical research. Accordingly, were this screening to become available to prospective parents, it should be implemented via a more robust model, whether that consists of unrestricted access for all parents or another model altogether. The results of this study contribute to continued ethical debate around acceptable models of practice as NIPT panels inevitably expand to include AOCs and beyond.

Acknowledgements We wish to acknowledge the study participants. We also wish to acknowledge Dr Hilary Bowman-Smart and Dr Molly Johnston for their assistance with survey content and structure. Lastly, we wish to acknowledge The Uehiro Foundation on Ethics and Education.

Contributors All authors contributed to this work. IM, CM and CD conceptualised and implemented the study. Analyses were undertaked and finalised by IM. Drafting of the manuscript was undertaken by IM. Review and editing were completed by all authors. IM is the author acting as the guarantor for this work.

Funding This study was supported by Australian Research Council (LP190100841).

Competing interests CM has received funding from the Australian Research Council for the project LP190100841, which includes partnership funding from Illumina and Victorian Clinical Genetic Services. Funding from Illumina is a potential conflict of interest.

Patient consent for publication Not applicable.

Ethics approval This study involves human participants and was approved by Monash University Human Research Ethics Committee (project number 29350). Participants gave informed consent to participate in the study before taking part.

Provenance and peer review Not commissioned; externally peer reviewed.

Data availability statement Data are available upon request.

ORCID iDs
India R Marks http://orcid.org/0000-0003-4770-8316
Katrien Devolder http://orcid.org/0000-0001-5022-4473

REFERENCES

1 Lo YM, Corbetta N, Chamberlain PF, *et al*. Presence of fetal DNA in maternal plasma and serum. *Lancet* 1997;350:485–7.
2 Dondorp W, de Wert G, Bombard Y, *et al*. Non-Invasive prenatal testing for aneuploidy and beyond: challenges of responsible innovation in prenatal screening. *Eur J Hum Genet* 2015;23:1438–50.
3 MacLeod R, Tibben A, Frontali M, *et al*. Recommendations for the predictive genetic test in Huntington's disease. *Clin Genet* 2013;83:221–31.

J Med Ethics: first published as 10.1136/jme-2023-109070 on 3 May 2023. Downloaded from http://jme.bmj.com/ on November 30, 2025 at Zanjan University of Medical Sciences. Protected by copyright, including for uses related to text and data mining, AI training, and similar technologies.

4 de Die-Smulders CEM, de Wert GMWR, Liebaers I, *et al*. Reproductive options for prospective parents in families with Huntington's disease: clinical, psychological and ethical reflections. *Hum Reprod Update* 2013;19:304–15.
5 Evers-Kiebooms G, Harper P, Zoeteweij MW. *Prenatal testing for late-onset neurogenetic diseases*. Oxford, United Kingdom: CRC Press LLC, n.d.: 1–220.
6 Duncan RE, Foddy B, Delatycki MB. Refusing to provide a prenatal test: can it ever be ethical? *BMJ* 2006;333:1066–8.
7 the Public Policy Committee of NSGC, Hercher L, Uhlmann WR, *et al*. Prenatal testing for adult-onset conditions: the position of the National Society of genetic counselors. *J Genet Counsel* 2016;25:1139–45.
8 de Jong A, de Wert GMWR. Prenatal screening: an ethical agenda for the near future. *Bioethics* 2015;29:46–55.
9 Laurie GT. In defence of ignorance: genetic information and the right not to know. *Eur J Health Law* 1999;6:119–32.
10 Bunnik EM, de Jong A, Nijsingh N, *et al*. The new genetics and informed consent: differentiating choice to preserve autonomy. *Bioethics* 2013;27:348–55.
11 Baig SS, Strong M, Rosser E, *et al*. 22 years of predictive testing for Huntington's disease: the experience of the UK Huntington's prediction Consortium. *Eur J Hum Genet* 2016;24:1515.
12 Duncan RE, Savulescu J, Gillam L, *et al*. An international survey of predictive genetic testing in children for adult onset conditions. *Genet Med* 2005;7:390–6.
13 Mand C, Gillam L, Delatycki MB, *et al*. Predictive genetic testing in minors for late-onset conditions: a chronological and analytical review of the ethical arguments. *J Med Ethics* 2012;38:519–24.
14 Duncan RE, Delatycki MB. Predictive genetic testing in young people for adult-onset conditions: where is the empirical evidence? *Clin Genet* 2006;69:8–16.
15 Verbeek INE, van Onzenoort-Bokken L, Zegers SHJ. Vulnerable child syndrome in everyday paediatric practice: a condition deserving attention and new perspectives. *Acta Paediatr* 2021;110:397–9.
16 Vears DF, Ayres S, Boyle J, *et al*. Human genetics Society of Australasia position statement: predictive and presymptomatic genetic testing in adults and children. *Twin Res Hum Genet* 2020;23:184–9.
17 Australian Government National Health and Medical Research Council. Medical genetic testing: information for health professionals. 2010. Available: https://www.nhmrc.gov.au/about-us/publications/medical-genetic-testing-information-health-professionals#block-views-block-file-attachments-content-block-1 [Accessed 11 Mar 2021].
18 Wade CH, Wilfond BS, McBride CM. Effects of genetic risk information on children's psychosocial wellbeing: a systematic review of the literature. *Genet Med* 2010;12:317–26.
19 Wakefield CE, Hanlon LV, Tucker KM, *et al*. The psychological impact of genetic information on children: a systematic review. *Genet Med* 2016;18:755–62.
20 Robertson S, Savulescu J. Is there a case in favour of predictive genetic testing in young children? *Bioethics* 2001;15:26–49.
21 Duncan RE, Gillam L, Savulescu J, *et al*. "you're one of US now": young people describe their experiences of predictive genetic testing for Huntington disease (HD) and familial adenomatous polyposis (FAP). *Am J Med Genet C Semin Med Genet* 2008;148C:47–55.
22 Taylor-Sands M, Bowman-Smart H. Non-Invasive prenatal testing for adult-onset conditions: reproductive choice and the welfare of the future child. *Mon L Rev* 2022;45:728–77.
23 Berkman BE. Refuting the right not to know. *J Health Care Law Policy* 2017;19:1–72.
24 Bowman-Smart H, Taylor-Sands M. Fetal information as shared information: using NIPT to test for adult-onset conditions. *Monash Bioeth Rev* 2021;39:82–102.
25 Begović D. Prenatal testing: does reproductive autonomy succeed in dispelling eugenic concerns? *Bioethics* 2019;33:958–64.
26 Turner PN. "Harm" and'Mill's harm principle. *Ethics* 2014;124:299–326.
27 Marks I, Devolder K, Bowman-Smart H, *et al*. Non-Invasive prenatal testing (NIPT) for adult-onset conditions: an examination of NIPT users. 2023.
28 Garrett JR, Lantos JD, Biesecker LG, *et al*. Rethinking the "open future" argument against predictive genetic testing of children. *Genetics in Medicine* 2019;21:2190–8.
29 Salloch S, Vollmann J, Schildmann J. Ethics by opinion Poll? the functions of attitudes research for normative deliberations in medical ethics. *J Med Ethics* 2014;40:597–602.
30 Bowman-Smart H, Savulescu J, Mand C, *et al*. "is it better not to know certain things?": views of women who have undergone non-invasive prenatal testing on its possible future applications. *J Med Ethics* 2019;45:231–8.
31 Millo T, Douiev L, Popper D, *et al*. Personalized prenatal genomic testing: couples' experience with choice regarding uncertain and adult-onset findings from chromosomal-microarray-analysis. *Prenat Diagn* 2021;41:376–83.
32 Borry P, Favaretto M, Batthyany A, *et al*. Noninvasive prenatal testing: a survey of young (future) parents in Flanders. *Personalized Medicine* 2018;15:35–43.
33 van Schendel RV, Dondorp WJ, Timmermans DRM, *et al*. NIPT-based screening for Down syndrome and beyond: what do pregnant women think? *Prenat Diagn* 2015;35:598–604.

J Med Ethics: first published as 10.1136/jme-2023-109070 on 3 May 2023. Downloaded from http://jme.bmj.com/ on November 30, 2025 at Zanjan University of Medical Sciences. Protected by copyright, including for uses related to text and data mining, AI training, and similar technologies.