

# Are prenatal genetic tests becoming too complex?

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As noninvasive prenatal testing becomes more advanced, questions of informed consent, clinical utility and ethical concerns become more complicated for clinicians, and more anxiety-provoking for parents,

according to the authors of an Ethics and Law article published online today by the *Medical Journal of Australia*.

Non-invasive prenatal testing (NIPT), introduced in 2010, was "revolutionary, with sensitivity, specificity and [detection rates](#) unmatched," according to the authors, led by Dr. Joseph Thomas, a senior specialist in maternal-fetal medicine at Mater Health Services in Brisbane.

"NIPT was found to achieve a detection rate for Down syndrome of 99.7%, with a false positive rate of 0.04%," Thomas and colleagues wrote.

However, as the technology advanced, some NIPT providers started to offer extended panels and low resolution whole genomic sequencing (WGS), including sex chromosome aneuploidies, recurrent microdeletions, subchromosomal deletions and duplications.

"This comes at a cost of a higher false positive rate and lower positive predictive value," Thomas and colleagues wrote.

"Moreover, the expanded panels and WGS NIPT raise issues of clinical utility and [ethical concerns](#)."

Ethical concerns include:

- the challenges in providing adequate information arising from the complexity of the tests—"From an ethical perspective, however, it is the understanding of information that is important, not merely that a person was given the legally required information";
- the risk of power imbalances and "normalization" of testing—"whereby a patient simply agrees because 'doctor knows

best' and, second, the impression that NIPT is a normal part of care, and therefore would be foolish to reject";

- anxiety resulting from complex and potentially unnecessary medical decisions—"higher levels of decisional regret among parents whose results identified variation of uncertain significance";
- the problem of screening for "normality" and genetic reductionism—"just because a genetic anomaly can be identified does not necessarily mean that it would be phenotypically expressed; similarly, detection of genes associated with adult onset disease does not necessarily equate to disease"; and
- the doctor's responsibility in determining which NIPT test is clinically indicated—"screening should be recommended or chosen only if there is likely to be a proportionate benefit, and there is no disproportionate burden."

Thomas and colleagues made the following recommendations:

- Informed consent is required for all NIPT tests, especially in the context of extended panels and WGS NIPT. Clinicians must understand the different abnormalities targeted by extended NIPT panels and be able to assess and communicate the clinical utility of screening in accordance with a particular patient's needs, desires and circumstances;
- If ordering WGS NIPT, given that there may be significant uncertainty as to the actual phenotypic or functional manifestation of a genetic variation in a particular child, the consent process should include helping to contextualize limitations and risks in the broader context of the human experience of risk and uncertainty;
- Genuine shared decision-making models can empower patient autonomy by helping them to understand the implications of their possible decisions in relation to their values. Moreover, decision

tools and algorithms that align a variety of scenarios with personal values can facilitate a high quality informed consent process;

- Higher resolution WGS NIPT should only be used for research purposes until we have robust data regarding its clinical utility.

**More information:** Joseph Thomas et al. Non-invasive prenatal testing: clinical utility and ethical concerns about recent advances, *Medical Journal of Australia* (2021). [DOI: 10.5694/mja2.50928](https://doi.org/10.5694/mja2.50928)

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