

Clinicians' Experiences of Explaining Prenatal Screening and Delivering Genetic Syndrome Diagnoses



Project aims and background

This project investigated healthcare professionals' experiences of explaining prenatal screening and delivering genetic syndrome diagnoses, and their perspectives on the new Prenatal Screening website (<https://prenatalscreening.org.au>) developed by Down Syndrome Queensland (DSQ) to support healthcare professionals in these areas.

The project developed from researchers at the Child Health Research Centre, University of Queensland, approaching DSQ to share their research findings on parents' negative experiences of their child being diagnosed with Down syndrome.

DSQ and our research team agreed that, to improve parents' experiences, we needed to better understand clinicians' perspectives. DSQ commissioned our team to conduct this research, which will be used to inform further resource development.

We conducted a survey and qualitative interviews with healthcare professionals across Australia.

- We surveyed 51 clinicians
- We interviewed 12 clinicians
 - 6 midwives, 4 GPs, 1 nurse, 1 neonatologist

In this report we describe key themes and findings.

Key challenges

Key barriers to patients accessing the NIPT are related to geographic location, health literacy, and financial resources:

- some patients in rural areas must travel long distances to receive healthcare, or they may be unaware of the NIPT, and so they delay seeking healthcare until later in their pregnancy
- the combination of decreasing availability of bulk-billed appointments and the expensive out-of-pocket cost for the NIPT present financial barriers

Misunderstandings of the NIPT and informed consent encountered by some clinicians include:

- the presumption that the NIPT does not require consent because it is non-invasive
- misinterpreting a high chance NIPT result as a diagnosis
- patients perceiving consent as having legal connotations and a sense of finality rather than being ongoing and changeable;
 - some clinicians opt for using 'choice' over 'consent'
- some clinicians involved in later stages of care find that patients are not fully informed about the NIPT that they consented to, e.g., they think the NIPT is a test to determine the baby's sex

Structural and logistical influences on prenatal screening:

- fragmented communication between midwives, GPs, and other clinicians can affect patient care and can cause confusion
 - e.g., pathology labs can use language inconsistently
- clinicians highlighted the benefits of continuity of care for both patient outcomes and their own confidence as clinicians
- quality communication and care requires sufficient time
 - clinicians navigate difficult time constraints of short appointments, having to explain a large amount of prenatal screening information alongside other pregnancy information, e.g., smoking, diet, etc.
 - pregnancy timelines also impact testing options

Communication strategies

Clinicians shared that they use a range of communication strategies for prenatal screening conversations, and for talking with patients who do not want non-invasive prenatal testing (NIPT) and patients who have unexpected results.

Clinicians communicating about prenatal screening aim to:

- continually ensure that the patient is informed, and aware of supports and the next steps
- follow a consistent process for communication
- build rapport efficiently and tailor communication and care to meet patients' needs, including cultural and religious sensitivity
- explain to patients that the NIPT is not diagnostic

Clinicians communicating with patients who do want the NIPT:

- take time to understand and address patients' misconceptions
- explain to patients that they have the right to abstain from the NIPT or ask for alternate care
- sometimes keep detailed documentation of a patient's decision to abstain from prenatal testing, to better inform the clinicians who will care for the patient in future

Clinicians communicating with patients who receive unexpected results:

- prefer to communicate results face-to-face with the patient, ideally in a private space, with their partner, and without interruptions
- view explaining the next steps as being critical for mentally preparing patients
- consider following up with a patient as being important – particularly when a patient is distressed and may not recall information talked about in the appointment

Misinterpreting the NIPT as a diagnostic test

When asked how they would respond to a patient choosing to terminate a high chance pregnancy, only ¼ of the 12 interviewed clinicians said that they would explain that the NIPT is a probability, not a diagnosis, and would offer the patient diagnostic testing.

Most clinicians stated that they would support the patient's decision to terminate and did not mention diagnostic testing. Some clinicians may have misinterpreted the question and assumed that the patient had a diagnostic test result; however, the lack of clarification-seeking remains problematic. This indicates that some clinicians are potentially misinterpreting the NIPT as a diagnostic tool.

“it's called non-invasive prenatal testing, which makes you think that it is a test, a diagnostic test. It's not called screening”

Clinicians' perspectives on the usage of 'risk'

Clinicians described mixed opinions on the usage of 'risk':

- some clinicians prefer using 'risk'
 - 'risk' is a longstanding medical term widely used by most medical professionals and pathology providers, and clinicians want to avoid creating misunderstandings
- some clinicians prefer using 'chance' or 'likelihood'
 - risk can be perceived as having negative connotations or being unnecessarily anxiety-provoking
 - there has been a shift away from using 'risk', towards use of inclusive language i.e., 'chance' or 'likelihood' as an alternative to 'risk'

Clinicians note that risk can be hard for patients to conceptualise and that risk ratios require informed interpretation.



Results from 51 survey respondents



Clinicians' confidence and comfortableness

Approximately 1 in 4 survey respondents did not feel confident or comfortable discussing prenatal screening, and only 1 in 2 felt comfortable delivering unexpected genetic diagnoses. Insights from the qualitative interviews contextualise these findings. Clinicians generally conflated high chance and unexpected results with bad news and shared that a key challenge of prenatal screening and genetic diagnosis conversations is managing patients' emotional reactions and distress.

“They're all conditions of bad news and sharing bad news.”

Clinicians expressed concerns over the limits of their knowledge of prenatal screening:

- clinicians find it challenging to keep their knowledge of prenatal screening up-to-date given that this is an area with evolving technological development and the NIPT results require nuanced understanding of positive predictive values for accurate interpretation
- clinicians are concerned about the accuracy of the information that they deliver, and accordingly prioritise the use of up-to-date information and resources
- challenges can be exacerbated if clinicians are rarely having conversations about prenatal screening in their practice

Views on the DSQ Prenatal Screening website

Among the minority of survey respondents who knew about DSQ's prenatal screening website, most viewed it as being a useful and engaging source of information for both them and their patients.

“Lots of information in one place.”

Perspectives on resources and referrals

Half of the survey respondents did not feel that they had adequate access to resources to aid their conversations with patients.

- **80% of survey respondents have not referred to DSQ** or other support services in the past, but **64% would consider referring** in the future
- most survey respondents were not aware of, or did not know enough about, these services to confidently refer
- resources from government bodies were favoured owing to their perceived lack of bias
 - there was concern that resources from non-government bodies could be directive, e.g., discourage pregnancy termination

Responses from the qualitative interviews show variation in the kinds of resources that clinicians prefer and how they seek out resources:

- Some clinicians prefer hard copy resources
 - patients can take the information home to read later
 - avoids patients accessing information online where certain websites could be outdated or inaccurate
- In contrast, other clinicians prefer online resources
 - perceived to have up-to-date information
- Clinicians vary in how they source information
 - conducting their own research
 - receiving new information from patients
 - relying on colleagues or their manager to stay informed
 - relying on external organisations to forward research and resources

Clinicians also identified a lack of training on delivering prenatal results, recommending that training be delivered through universities but also potentially through learning modules, refresher courses, workshops/webinars, hospital education sessions, and conferences.

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Authors	
Emma Cooke	e.cooke@uq.edu.au
Mark Anderson	mark.anderson@uq.edu.au
Grace Langdon	g.langdon@uq.net.au
Madeline Feltham	m.feltham@uq.net.au
Sally Staton	s.staton@uq.edu.au
Laetitia Coles	l.coles@uq.edu.au
Karen Thorpe	k.thorpe@uq.edu.au
Jasneek Chawla	j.chawla@uq.edu.au

For further details contact:

The University of Queensland
kidssleep@uq.edu.au
uq.edu.au

