

What genetic testing information to expect when you're expecting

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Credit: Murdoch Childrens Research Institute

Experts have recommended greater support and choice of information for pregnant women who decide to have genetic testing.

The latest research from the Murdoch Children's Research Institute (MCRI), published in the *European Journal of Human Genetics*, has shown a divide in the amount of [genetic information](#) they want to receive from their prenatal [test](#). Current practice does not provide parents with such a [choice](#).

At a time when prenatal testing is becoming more widespread, there has

been little research to date about the amount of fetal information pregnant women might want.

A method used widely now for testing the fetal sample, chromosome microarray analysis, means we can obtain a lot more information about small variations in the DNA.

There are people who like to have all the information they can about their pregnancy, even if this information is uncertain. For other people, such uncertainty is difficult to deal with, and they would rather not know.

The [GaP study](#) is about letting pregnant women (and partners) choose how much information to receive. The options are 'targeted', where only the information that will affect health is provided, or 'extended', where all information, even the uncertain aspects, is provided. A decision aid has been produced to help understand the tests and choices available.

111 women participated in the GaP study from seven antenatal clinics in Victoria. At least two days before their procedure they were counselled and provided with a decision aid. This decision aid presented two options; to receive all genetic information or to receive only information that was known to affect function.

Researchers found that 60 percent chose to receive all information and 40 percent chose the limited amount, a clear demarcation in choice. There were no adverse psychological affects as measured by surveys before and after the testing.

The study was done with [pregnant women](#) carrying a baby with no visible abnormality on ultrasound, but who were having [genetic testing](#) (chromosome microarray analysis) following a prenatal diagnosis procedure, either chorionic villus sampling or amniocentesis, done for

other reasons.

Lead author, MCRI Genetics Group Leader, Professor Jane Halliday said "There will always be uncertain or unknown variation in chromosomal material detected by genetic testing, suggesting probability of or susceptibility to an adverse health outcome."

"Such uncertainty can create confusion, conflict, anxiety about how to manage the situation, while certainty of outcome minimises these emotions."

"In a medical era that promotes patient-centred decision-making, offering individualised choice about the scope and depth of genetic analysis, whether it be in pregnancy or later, seems more important than ever."

Experts have recommended the use of a decision tool, such as the decision-aid, along with pre-test genetic counselling, to better facilitate greater understanding of all the test-related genetic information.

Senior author Professor David Amor said, "In order to provide choice in a clinical setting, the testing laboratories would have to agree to the two-tier approach and manage the responsibility of not providing susceptibility or uncertain genetic information, if that has been the request of the patient."

"Such changes will require significant resource for the laboratory, and professional bodies such as RANZCOG/ HGSA will need to develop policy to support and facilitate this clinical pathway. In addition, antenatal clinics will have to introduce a different model for counselling which allows for specific choice."

Emily's story

Emily Piggot took part in the GaP Study when she was pregnant with her second child Edith, who is nearly two and a half.

At 40, Emily was at a higher risk of complications in pregnancy so she and her partner decided they wanted to be more informed so they could make the best decision about their pregnancy.

Emily said initially she was fascinated knowing she could potentially have access to a lot of genetic information but then decided they wanted only enough [information](#) that could influence a decision on whether or not to continue the pregnancy and nothing further.

"I thought that if I learnt about a condition that may, or may not develop I would have a lifelong burden about whether to tell her or not.

"It's kind of like knowing that your kids are doing something naughty but not particularly bad, sometimes you'd just rather not know about it," Emily said.

Provided by Murdoch Childrens Research Institute

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