Decision Aid

Choosing between extended and targeted prenatal information
Welcome to the GaP study
(Genomics and Pregnancy)

Thank you for your interest in the GaP study. You have been invited to participate based on your decision to have a prenatal diagnostic test; i.e. chorionic villus sample (CVS) or amniocentesis.

As a participant of this study, you will be given the opportunity to choose how much genetic information to receive from this test. In current practice, women (and partners) are not offered this choice.

You will be asked to choose between receiving:

• **Extended** information, and
• **Targeted** information.

This booklet is designed to help you (and your partner) understand the key differences between these two types of information, and to help you choose which one is right for you.

The choice you are about to make will determine how much, and what sort of genetic information will be reported about your pregnancy.
What does testing involve?

Prenatal tests are used to look at the chromosomes and DNA of a developing baby. There are two different laboratory tests.

**Test 1  First rapid test;** results in 1-2 working days

**Test 2  Second more advanced test;** results in 10-14 working days

What can the first rapid test detect?

The *first rapid test* produces results in 24-48 hours. This test is called the *FISH test*. It can detect some of the common chromosome problems found during pregnancy. In most cases, nothing will be found at this initial stage of testing. In some cases, the following may be detected:

- Down syndrome (An extra copy of chromosome 21)
- Edwards syndrome (An extra copy of chromosome 18)
- Patau syndrome (An extra copy of chromosome 13)
- Turner syndrome (females with only one copy of the X chromosome)
- Klinefelter syndrome (males with an extra copy of the X chromosome)

More information about *FISH* is available in the *Fact Sheet* (page 13)

Your managing doctor or genetic counsellor will notify you of the result.

- If any one of the above conditions is found by the first rapid test, you will be able to discuss your options and further testing with your care provider.

- If *no* chromosome changes are found, the laboratory will proceed with the second more advanced test.
What can the second more advanced test detect?

The second more advanced test produces results in 10-14 working days. It is called a chromosomal microarray. It looks at the DNA more closely to detect if there are other changes (variations) that could have an impact on the health and/or development of the baby.

More information is available in the Fact Sheet (page 13).

In most cases, no genetic variations are found. In some cases, this test will find:

- A variant known to affect health/development
- A variant with an unknown or uncertain affect on health/development

On average, out of 100 women who have this second more advanced test:

95 will have a result showing no genetic variation; i.e. ‘normal’ result

3 will have a result showing a genetic variation known to impact on health and/or development; i.e. ‘abnormal’ result

2 will have a result showing a genetic variation with unknown/uncertain outcome
What do unknown/uncertain results mean?

**Genetic variations with unknown/ uncertain outcome have either:**

i) never been seen before; their impact on health and/or development is ‘unknown’

or

ii) have been seen before, but their impact on health and/or development is ‘uncertain’

In some cases, both parents can have a blood test to determine if they carry the same unknown/uncertain variant found by the prenatal test. If one of the parents has the same variant, and their health is not affected, it is likely that the health of the developing baby will not be affected either.

There is still a lot to learn about genes and how they work. We are all born with some genetic variations; yet, we don’t always know if they affect health or not.

When such variations are found during pregnancy, it can be hard to predict what the outcome will be. They may or may not have an impact on growth and development.

These results can be difficult to deal with, but your healthcare provider will be available to support you and help you understand what these results might mean.
How much information will I receive?

As a participant in the GaP study, you are asked to make a decision about receiving \textit{extended} or \textit{targeted} information from your prenatal diagnostic test.

\textbf{You can choose:}

- \textbf{to know} about the variants of unknown/uncertain significance, and therefore to receive the \textit{extended} set of information.

- \textbf{to not know} about the variants of unknown/uncertain significance, and therefore to receive the \textit{targeted} set of information.

\textbf{Example:} You decide to participate in the GaP study. Your test finds a genetic variant of unknown/uncertain significance. If you selected to receive:

- \textit{extended} information, you \textbf{will} be told about these results.

- \textit{targeted} information, you \textbf{will not} be told about these results.
Prenatal testing summary

**CVS or amniocentesis**

**First rapid test**
Results available in 24-48 hours
- No chromosome abnormality
- Down syndrome (Trisomy 21)
- Edwards syndrome (Trisomy 18)
- Patau syndrome (Trisomy 13)
- Turner & Klinefelter syndrome

**Second more advanced test**
Results available in 10-14 working days

<table>
<thead>
<tr>
<th></th>
<th>Extended information</th>
<th>Targeted information</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>No variation detected</strong></td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td><strong>Variation with unknown/uncertain outcome</strong></td>
<td>✓</td>
<td>✗</td>
</tr>
<tr>
<td><strong>Variation with known abnormal outcome</strong></td>
<td>✓</td>
<td>✓</td>
</tr>
</tbody>
</table>

More consideration and discussion about what this means for you

✓ you will receive this result
✗ you will not receive this result
Need more information?

The next pages provide:

• A summary of the differences between each type of information
• A worksheet to help you make a decision
• Practical considerations
• Fact Sheet
• Glossary
What does this all mean?

Prenatal testing can provide information to parents about the health of the baby. Sometimes, prenatal testing may not provide certain answers; some results can be unclear and hard to deal with.

<table>
<thead>
<tr>
<th>Advantages</th>
<th>Disadvantages</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Extended information</strong></td>
<td>All the available information about the genetic variations found is reported, which can be reassuring.</td>
</tr>
<tr>
<td><strong>Targeted information</strong></td>
<td>Only known variations are reported, so there is less uncertainty and confusion to deal with.</td>
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</tbody>
</table>

*Genetic counselling is available to support decision making when unclear results are found.
Worksheet: Extended or Targeted information?

This page is designed to help you decide whether extended or targeted information is best for you.

Read the statements below and put a tick in the box that corresponds to your answer.

<table>
<thead>
<tr>
<th>Agree</th>
<th>Disagree</th>
</tr>
</thead>
<tbody>
<tr>
<td>I don’t want to know about variations with unknown/ uncertain outcome</td>
<td></td>
</tr>
<tr>
<td>I want as much information about my baby as possible</td>
<td></td>
</tr>
<tr>
<td>Being told about variations with unknown/ uncertain outcome would not be confusing to me</td>
<td></td>
</tr>
<tr>
<td>I don’t want all available information to be reported to me</td>
<td></td>
</tr>
<tr>
<td>I would be OK knowing unclear information is not being reported to me</td>
<td></td>
</tr>
</tbody>
</table>

Which way are you leaning in your decision?

Add up the ticks in each column.

- How many under ‘Agree’?
- How many under ‘Disagree’?

If you have more under ‘Agree’, you are likely to prefer **targeted information**

If you have more under ‘Disagree’, you are likely to prefer **extended information**
Other practical considerations

How much time do I have to make a decision?

You have until the day of the CVS or amniocentesis procedure to decide how much information you want from this test. **Make sure you indicate your choice on Survey I.**

Is there a cost involved?

There is no additional cost involved to participate in this study.

How will I find out about the results?

Your managing clinician (e.g. GP, obstetrician, midwife, medical geneticist or genetic counsellor) will contact you once he/she receives your results from the laboratory. Together, you will be able to discuss the possible implications of the results.

Rapid test results can be expected between 24-48 hours; results from the second, advanced test will be available in 10-14 working days.
Where can I get more information?

The GaP research team will be able to answer your questions about the study:

by phone: (03) 9936 6766, or
by email: gap.study@mcri.edu.au
web: www.mcri.edu.au/research/research-projects/gap-study/

Where can I get more information about prenatal testing?

Your managing clinician (e.g. GP, obstetrician, midwife, medical geneticist or genetic counsellor) who has offered you the prenatal diagnostic test, will be able to answer your questions about prenatal testing.

More information can also be found on the Raising Children Network website: http://www.raisingchildren.net.au/articles/disability_antenatal_tests.html


Information & support networks


by phone: (03) 8341 6315
by email: info@gsnv.org.au
Fact Sheet
More information on the two tests

**First rapid test: the FISH test**

FISH stands for fluorescence in situ hybridization. Using fluorescent dye, this test can show whether or not the pregnancy has the right number of chromosomes. Having too many (or not enough) chromosomes can affect health and/or development. This test examines chromosomes 13, 18, 21, X and Y.

For example, the FISH test can determine if there are 3 copies of the chromosome 21 (instead of 2). This would indicate Down syndrome.

**Second more advanced test: Chromosomal microarray**

The chromosomal microarray (CMA) is a new test that allows laboratory technicians to ‘zoom in’ on the chromosomes and analyse the DNA content very carefully.

CMA can detect whether chromosomes carry small variants, such as additions or deletions of DNA. These variants are too small to be detected using traditional tests. Some of these variants are known to affect health, while others may (or may not) have any impact (i.e. genetic variation with unknown or uncertain outcome).
## Glossary

<table>
<thead>
<tr>
<th>Term</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Amniocentesis</strong></td>
<td>A diagnostic test (from 15 weeks) where a small sample of amniotic fluid is collected for testing.</td>
</tr>
<tr>
<td><strong>Chorionic Villus Sampling (CVS)</strong></td>
<td>A diagnostic test (done between 11-13 weeks) where a sample of the placenta is collected for testing.</td>
</tr>
<tr>
<td><strong>Chromosomes</strong></td>
<td>An organised package of DNA. Humans have 23 pairs of chromosomes. Extra or missing chromosomes can affect health &amp; development.</td>
</tr>
<tr>
<td><strong>DNA</strong></td>
<td>Stands for deoxyribonucleic acid. It is the genetic material that carries the instructions for all living things. A useful analogy: DNA (words) – genes (sentence) – chromosomes (chapter) – genome (book).</td>
</tr>
<tr>
<td><strong>DNA variants</strong></td>
<td>Small additions or deletions in the DNA.</td>
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<tr>
<td><strong>Genetic counsellor</strong></td>
<td>A health professional with specialised training in genetics and counselling. They provide information and support to individuals and families who have, or are at risk for, chromosomal disorders and genetic conditions.</td>
</tr>
<tr>
<td><strong>Medical geneticist</strong></td>
<td>Doctor who specialises in the diagnosis, management and treatment of genetic conditions.</td>
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