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AMNIOCENTESIS/CVS

This letter contains information which we ask you to read before your appointment. At the consultation, you can discuss with Dr Carmody any issues or questions you might have. You will then be asked to consider this information before signing a consent form and undergoing the procedure.

For directions and parking information for both our clinics, visit our website at www.fetalmedicine.com.au Go to Patient Details, then Contact Details.

In order to obtain a Medicare refund, you must have a referral for this scan and procedure. Please bring this with you on the day, unless you or your doctor have organised to fax or send it to us prior to the appointment. Dr Carmody must also sight written evidence of your blood group. Your referring Doctor may have already written this on your referral.

For medico-legal reasons, children under six years of age are NOT permitted in the ultrasound rooms. If you have to bring your child, it is essential that another adult accompanies you, so that your child has supervision in the waiting room.

This appointment will take approximately 60 minutes for a singleton, or 90 minutes for a twin pregnancy. We do our best to run on time, however situations outside of our control will cause delays in the scheduling eg when a baby is in a difficult position for scanning, or an anbormality is found in a pregnancy etc. These sorts of issues do mean that you might be here longer than the allocated time.

You will receive a USB, photos, and a copy of your report for your keepsake folder. Please note that the quality and number of images provided is reliant on several factors, including the patient's BMI, the fetal position and the amount of amniotic fluid.

We will confirm your appointment two to three working days before the scheduled date. If you don't hear from us, please ring to confirm. If for some reason you are unable to keep your appointment, **please notify us as soon as possible.**

The fee for an amniocentesis (this includes the initial consultation, an ultrasound and the amniocentesis) is $405.00 and is **payable on consultation**. The fee for a chorionic villus sampling (this includes the initial consultation, an ultrasound and the CVS) is $425.00, and is **payable on consultation.** Multiple pregnancies attract an additional cost.

PLEASE NOTE

**You will also receive an account from either S&N or QML for the pathology.
This account is partially rebatable through Medicare.**

Please direct any queries regarding the pathology expense or account to the relevant provider i.e. S & N or QML.

It is not necessary to have a full bladder. After the procedure, it is advisable that you have a "restful day". It is not necessary for you to go to bed, but you should avoid unnecessary activity. It is OK to drive a motor vehicle.

You may experience some mild lower abdominal cramping. This should not be of concern. However if you experience any bleeding, loss of fluid through the vagina, or severe cramping, lower abdominal pains, you should contact either myself or your referring Obstetrician. Most women who experience these symptoms DO NOT go on to miscarry.

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The results are faxed to the Wesley rooms from 5.30pm on the next business day following your procedure. We will phone you from this office as soon as we have these results.

AMNIOCENTESIS AND CVS

Your questions answered

The issues surrounding amniocentesis and Chorionic Villi Sampling (CVS) are complex. This leaflet addresses the more common concerns. If you have further questions, please feel free to ring, or ask our clinical staff when you attend for your appointment. If, after discussion, you elect not to proceed, there is no pressure to do so.

Who decides if I should have an amniocentesis or CVS?
You do. Your obstetrician or our staff can give you information to help you decide, but the final decision is yours.

Why should I consider having these tests?
The most common reason is to determine whether the baby has Down syndrome. Other reasons include a suspicious scan, a previous baby with a problem or a family history of certain conditions.

The only fetal abnormality that increases with maternal age is that the fetus could have the wrong number of chromosomes. If you have a normal amniocentesis or CVS, and a normal scan at 18-20 weeks, your chances of having a normal, healthy baby would be high.

Different people accept different levels and types of risks. Some say they couldn't risk a miscarriage or undergo a termination no matter what. Others say they couldn't lay the burden of a Down syndrome child on their other children, either during their childhood or after the parents die. Other people say they probably wouldn't undergo a termination, but would like to know how to prepare themselves if necessary.

What is my risk for having a baby with Down syndrome?
There are various ways of assessing the risk. The charts used later in this leaflet are based on maternal age at conception, and looks at the risk of having a live-born Down syndrome baby. They also assume no previous or family history of problems like Down syndrome. The charts are quoted later as they also include other figures that need explaining.

What are the risks of having an amniocentesis or CVS?
The main concern is the risk of miscarriage. Across the literature, the accepted rate for an amniocentesis is around 1 miscarriage in 1000 procedures. We have a considerably lower miscarriage rate. There is no way of predicting who will miscarry. For a CVS, 1 miscarriage in 500 would be considered attributable to this procedure.

Comparing your risk from the table with these risks may help you decide whether to have the procedure, although we understand that there is more to the decision than just numbers. However, if the risk for finding something bad is higher than the risk of doing something bad, you may feel more comfortable about having the procedure. Rarely, other problems may occur. The procedure or the laboratory testing may not be successful.

It is important to understand that amniocentesis looks for specific things such as Down syndrome. It does not rule out all abnormalities. It is important to have a morphology scan at 18-20 weeks to exclude many other problems. Think of the chromosomes as building plans, and the baby's development as the building process. Amniocentesis ensures that the plans are correct, and the 18-20 week scan is like the building inspector checking up part way through the building process.

Unfortunately, there is no test that can exclude mental retardation from other causes, including cerebral palsy.

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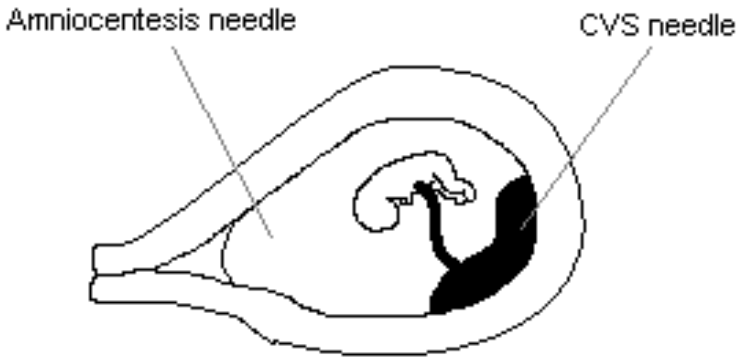
When is the procedure done?
A CVS is performed at 12 weeks. An amniocentesis is routinely performed between 14-16 weeks, although it can be done later for various reasons.

How is the procedure performed?
After consultation, a preliminary scan is made to assess the baby as far as possible and to check the dates. The best place for the needle is marked on the skin. This depends on the position of the baby and the placenta.

The mother's abdomen is cleaned with antiseptic and a local anaesthetic is usually given. This will sting briefly and then go numb. Most ladies say that this is the worst part.

The main needle is carefully inserted whilst scanning to avoid the baby. The lining of the uterus cannot be numbed without potentially compromising the baby. Some discomfort will be felt as the main needle passes through this lining, although it doesn't worry most women.



Twenty mls of fluid is withdrawn. This takes a minute or two due to the narrow gauge of the needle. The needle is withdrawn and the baby is then rescanned to ensure that no problems have occurred.

The whole procedure involving needles usually takes around five minutes, although we may need to pause if the baby approaches the needle.

Why is 20 mls of fluid needed?
The fluid contains a small number of skin cells that have washed off the baby. These are what the laboratory actually cultures to check the chromosomes. If we take less than 10 mls, the laboratory may not have sufficient cells.

The amount of fluid varies with gestation from person to person, but there is usually around 350-400 mls at the time of the procedure. The baby makes up the difference in a few days. Most women say that the procedure was not as bad as they imagined.

What is Chorionic Villi Sampling (CVS)?
This yields similar results to amniocentesis, but the technique is different. After the local anaesthetic, a different type of needle is inserted through the abdominal wall. Small snips of tissue are biopsied from the back of the placenta.

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The advantages of CVS are that it can be done from 12 weeks and it may be more appropriate for specific inherited disorders such as Cystic Fibrosis. The disadvantages include a higher incidence of bleeding and miscarriage and more discomfort. There is also a 4% chance that the laboratory will return 'mixed results'. If this happens, an amniocentesis would be recommended.

What happens afterwards?
Most women are able to continue their normal activities. Extreme activities should be avoided for a day or two. A few women experience a period-type pain almost immediately after the procedure. This is not related to miscarriage. It is the muscle of the uterus reacting to the needle. It should settle fairly quickly.

What should I look for?
1% of women who have had an amniocentesis or CVS will experience a small amniotic fluid loss (leak) or bleeding. This is usually of no concern and will normally cease within 48 hours. If this happens to you please:

- Rest in bed for 48 hours
- Do not have a bath (showering is fine)
- Do not have sex
- Call Dr Carmody's rooms on 3371 4933. We will book an appointment for a scan 48-72 hours after the procedure.

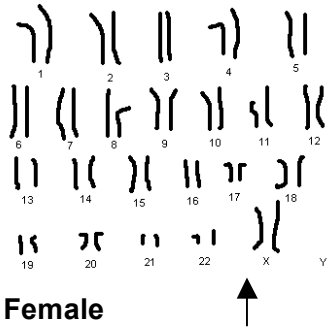
If you experience severe pain, blood loss or large gush of fluid out of our office hours, please proceed to your closest hospital – they will contact Dr Carmody. If there is no leak or bleeding within one hour after the procedure, you can resume life as normal. Paracetamol is helpful for mild uterine cramping.

Bleeding or fluid seepage is far more likely to settle than not, as long as you rest. In extreme cases, the problem may progress to a miscarriage.

A scan at the time of the problem is unnecessary, as it would only show what we already knew. Generally, a scan in a day or two would be suggested, to see if the situation is settling.

The miscarriage rate of 1 in 200 includes those that would have happened anyway. If a miscarriage is going to happen, the signs are most likely to appear the same day. If 48 hours pass with no signs, it is highly unlikely that you will miscarry. Any problems after this time are probably not due to the procedure.

Why does Down syndrome happen?
Almost all cells in the body contain 46 chromosomes. There are 23 pairs: 22 general pairs (numbered from longest to shortest), and 2 sex chromosomes. XX is female and XY is male. The pattern of a normal female is shown below:



Female

The baby receives one of each chromosome from each parent. Just before ovulation, the egg divides and changes from having 23 pairs of chromosomes into having 23 single chromosomes. These, when joined by 23 from the sperm, combine to return the number of chromosomes to 46.

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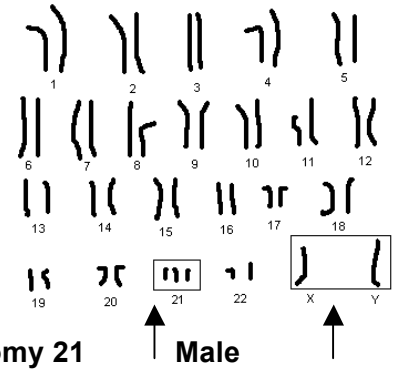
The father's age is not a concern as sperm regenerate every 40 days or so. However, women are born with all of their eggs. As the eggs get older, the bonds between the pairs get stickier. When the egg divides, the pairs may not separate cleanly. The entire pair may be kept or discarded. This is how some babies finish up with one more or one less chromosome and why it is more likely to occur as women get older. The viability depends on which pair is involved. Most of the unusual combinations are not viable and miscarry before 12 weeks.

Down syndrome is specifically an extra chromosome number 21, i.e. 3 x 21's. The technical term is Trisomy 21, (try-so-me 21). This is derived from tri-, meaning three and –somy, meaning chromosome. Trisomy 21 accounts for around half of all babies with an unusual combination of chromosome that have survived to 13 weeks.



Trisomy 21 **Male**

Although Down syndrome is more likely to occur as women get older, it can happen to younger women sporadically for reasons that we don't understand. In fact, around 80% of Down syndrome babies are now born to younger women. This is partly because older women are offered amniocentesis and the option of termination, but mainly because younger women are having most of the babies.

How accurate is the test for Down syndrome?
The amniocentesis results are virtually 100% accurate, as the laboratory are looking directly at the chromosomes. The most commonly reported cause for incorrect results is writing the wrong name on the test tube. Dr. Carmody will ask you to check this.

What else will the test find?
Down syndrome babies are the 'watershed', or the best of the survivors with unusual numbers of chromosomes. Babies with combinations that are worse tend not to survive. They usually miscarry before term, however they may live for up to a few months. The most common combinations in the group are Trisomies 13 and 18.

Babies with combinations which are better than Down syndrome involve 1 or 3 sex chromosomes. These babies do not have 'confused sex', but may have other problems. The actual manifestation depends on the combination.

With all of these combinations, there are varying degrees of severity. This cannot be ascertained until after the child grows and develops. For instance, no one can predict whether a Down syndrome baby will have mild or severe mental retardation.

What about Spina Bifida?
This is a hole in the spine and is one of a group of problems called neural tube defects.
While the amniotic fluid is in the laboratory, they measure a substance called Alpha Feto Protein , or AFP. If the level is normal, this would make Spina Bifida unlikely. An elevated level of AFP in the amniotic fluid would detect 97% of cases of Spina Bifida.

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How long will the results take?
The AFP results are usually through in a few days. The final amniocentesis results usually take around 10-14 days, depending on the gestation and how much the cells have to be cultured. We will not disclose the sex of the baby unless you ask.

In most cases a 24 hour result called Fluorescent In-Situ Hybridization, or FISH, which counts chromosomes 13 and 18 (for the ones which may survive this far into the pregnancy but won't survive in the long run), 21 (for Down syndrome) and the sex chromosomes is available. These account for over 90% of any bad results. FISH is accurate for what it checks for, but it cannot exclude everything.
Just occasionally, there may not be enough skin cells to perform both tests. In this case, the laboratory will skip the FISH and proceed with the main test.

We will ring you with the results as they come in, unless your doctor prefers to do this. A copy will be sent to your doctor.

I'm not keen on having an amniocentesis or CVS. What are the alternatives?

Unfortunately, the only way to be absolutely certain about things like Down syndrome is to have an amniocentesis or CVS.

If you wish, there are other tests available which you may find helpful. However, these are not as accurate.

At 11-14 weeks, we can perform a special ultrasound check for Nuchal Translucency, or puffy skin at the back of the neck. If this is found beyond a certain limit for a given gestation, it would raise our level of suspicion and we would recommend an amniocentesis. However, not all babies with Down syndrome have this puffy skin. That is, many would not be detected. The detection rate in the literature varies somewhat.

At 18 weeks, the normal 'Morphology Scan' can be performed. This may detect 'markers' or indicators that would make us suspicious, and we would recommend an amniocentesis. However, some babies with Down syndrome look totally normal on scan. Again, the detection rate in the literature varies.

You may be prepared to accept the above tests, but they cannot guarantee that your baby does not have Down syndrome. There are differing opinions as the effectiveness of combining the results of the different tests.

Unfortunately, the bottom line is that the only YES/NO tests available for Down syndrome are amniocentesis or CVS.

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Risk Chart

This chart is based on maternal age at conception and looks at the risk of having a live-born Down syndrome baby. The second column is the risk of having a live born baby with the wrong number of chromosomes, including Down syndrome.

Age	Downs	All
20	1:1667	1:526
21	1:1667	1:526
22	1:1429	1:500
23	1:1429	1:500
24	1:1250	1:476
25	1:1250	1:476
26	1:1176	1:476
27	1:1111	1:455
28	1:1053	1:435
29	1:1000	1:417
30	1:952	1:384
31	1:909	1:384
32	1:769	1:323
33	1:625	1:286
34	1:500	1:238
35	1:385	1:192
36	1:294	1:156
37	1:227	1:127
38	1:175	1:102
39	1:137	1:83
40	1:106	1:66
41	1:82	1:53
42	1:64	1:42
43	1:50	1:33
44	1:38	1:26
45	1:30	1:21
46	1:23	1:16
47	1:18	1:13
48	1:14	1:10
49	1:11	1:8