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GP’s role

• Identify opportunities for pre-pregnancy counselling.
• Collect relevant family history.
• Assess whether information in the family history places the current pregnancy at increased risk.
• Consider carrier tests for those from specific ethnic groups.
• Provide information about screening and diagnostic tests during pregnancy.
• If necessary, refer on to appropriate Genetics Services and/or support groups.
• Discuss peri-conceptual folic acid supplementation, the implications of drug and medication use during pregnancy, and other lifestyle modifications, eg smoking, drugs, alcohol.
• Provide information about infectious diseases during pregnancy.

Counselling before and during pregnancy

• If possible, pre-pregnancy counselling is advised and should include:
  > Collection of relevant family history (see ‘Collecting the family history’ below, and Family history).
  > Recommending periconceptional folic acid supplementation (see ‘Folic acid before and during pregnancy’ below).
  > Provision of information about screening and diagnostic tests during pregnancy (see ‘Types of prenatal tests’ below).
  > Consideration of carrier tests for those from specific ethnic groups, eg thalassaemia, sickle cell screening and Tay-Sachs disease (see Haemoglobinopathies).
  > Assessment of drug and medication use and implications for pregnancy.
  > Discussion of lifestyle changes, eg alcohol and smoking cessation during pregnancy and changes to diet.
  > Provision of information about infectious diseases:
    – Rubella vaccination status
    – Varicella antibody status and immunization if non-immune
    – Discussion about listeria infection and toxoplasmosis
  > Provision of information about pre-implantation genetic diagnosis (PGD), for couples who are known carriers of a genetic condition and/or couples undergoing IVF (see ‘Pre-implantation genetic diagnosis’).
  > Where couples are known carriers of a genetic condition, refer to Genetics Services.
Collecting the family history

- See *Genetics in practice* for further information.
- The family history of the woman and her partner should be collected regarding:
  - Inherited conditions, eg cystic fibrosis, fragile X syndrome, Duchenne muscular dystrophy
  - Down syndrome and other chromosomal abnormalities
  - Birth defects, eg spina bifida, cleft lip/palate, cardiac defects
  - Intellectual disability
  - Recurrent miscarriage
  - Unexplained perinatal deaths
  - Consanguinity (see *Genetics in practice*)
  - Ethnic background

Folic acid and pregnancy

- About 1 in every 500 pregnancies is affected by a neural tube defect.
- Research has shown that 70% of cases of neural tube defects (spina bifida, anencephaly, cleft lip with or without cleft palate) can be prevented by increasing the intake of folic acid prior to, and during early pregnancy.
- Folate is a B group vitamin found in leafy green vegetables, wholegrain breads, cereals and legumes. It is also available in tablet forms as folic acid.

**Recommendations about folic acid in pregnancy**

**Women at population risk for neural tube defects**

- Women planning a pregnancy should take supplementary folic acid, 0.5mg [500μg] folic acid tablet or multivitamin appropriate for use in pregnancy and containing at least 0.4 mg [400μg] of folic acid) every day for at least one month prior to possible conception and continued for the first three months of pregnancy.
- As many pregnancies are unplanned, all women of reproductive age should consider taking supplementary folic acid or a folate-rich diet.
- Folic acid tablets and multivitamins containing at least 0.4mg [400μg] folic acid are available from chemists, health food stores and some supermarkets.

**Women at increased risk for neural tube defects**

- Women are at higher risk of having a baby with a neural tube defect if:
  - They have had a baby with spina bifida, anencephaly or other neural tube defects
  - They themselves have had a neural tube defect
  - They are on certain medications for epilepsy
  - They have a close relative who has had a neural tube defect.
- These women should take supplementary folic acid every day for at least one month prior to possible conception and continued for the first three months of pregnancy. The dose recommended is usually 5mg [5000μg].

**Important points about folic acid**

- Women taking drugs to control epilepsy or seizures should ask their doctor whether they should increase the dose of folic acid to 5mg daily. However, specific evidence is limited in this area.
- Women planning to take multivitamins to provide folic acid supplementation should check with their pharmacist or doctor whether the multivitamin dose they are planning to use contains amounts of all the other vitamins/minerals that are safe for pregnancy, as well as providing the right amount of folic acid.
Assessing risk factors in pregnancy

**Down syndrome and other chromosomal abnormalities**

- All women are at risk of having a baby with a chromosomal abnormality, the most common being Down syndrome.
- The risk of Down syndrome increases with maternal age, as illustrated in Figure 1.

![Figure 1. Maternal age and risk of liveborn baby with Down syndrome](image)

**The effect of maternal age on screening tests for Down syndrome**

- Screening tests give a risk figure for Down syndrome that modifies the risk based on maternal age alone.
- The detection rate depends on the type of test.
- The detection rate and probability of an increased risk result increases with maternal age as the calculations of risk usually include the woman’s age.
- It is important that the woman/couple understands that screening tests will not identify all pregnancies with Down syndrome and that an increased risk result will require further clarification. The result from a chorionic villus sampling or amniocentesis will most likely still be normal.
- Screening tests should be offered to all pregnant women.

**Factors that increase the risk of having a baby with Down syndrome and other chromosome abnormalities**

- Maternal age.
- A previous pregnancy with a chromosome trisomy.
- An increased risk result on a screening test.
- The presence of soft signs of Down syndrome or other fetal anomalies during ultrasound examination.
- A parent carrying a chromosome rearrangement, eg a translocation involving chromosome 21 may increase the risk for Down syndrome. See Chromosomal conditions.
**Neural tube defects**

- The risk of a baby having a neural tube defect:
  - Is approximately 0.2%, but is higher if there is a past or family history of the condition
  - Does not increase with maternal age
  - Is increased for women taking certain anticonvulsants. Advice regarding risk is available from drug information services in obstetric hospitals or from Genetics Services

**Genetic conditions and birth defects**

- A family history of a birth defect (e.g., congenital heart defect), inborn error of metabolism (e.g., phenylketonuria) or other inherited conditions (e.g., thalassaemia) may indicate an increased risk of that condition.
- A family history should be collected from the woman and her partner. If the woman is concerned or there is a significant history they should be referred to Genetics Services for further risk assessment, preferably prior to conception.

### Types of prenatal tests

- **Screening tests** determine if the baby has an increased risk of having a particular problem such as Down syndrome or a neural tube defect. They are not diagnostic and an increased risk result does not mean the baby will definitely be affected.
  - Prenatal screening tests include:
    - Ultrasound
    - Early pregnancy (first trimester) screening: nuchal translucency ultrasound together with testing of the mother’s blood
    - Second trimester screening: testing the mother’s blood (maternal serum screening)
  - Prenatal screening tests should not be considered routine, but rather offered as a choice to women.

- **Diagnostic tests** determine if the baby has, or will develop after birth, a genetic condition. Sampling procedures to obtain cells for chromosome analysis or specific genetic tests are invasive.
  - Prenatal diagnostic tests include:
    - Ultrasound
    - Chorionic villus sampling (CVS)
    - Amniocentesis
  - Prenatal diagnostic tests should not be considered routine, but rather offered as a choice to women.
Figure 2. Prenatal screening and diagnostic tests offered

The flow chart shows the possible pathways a woman may take with respect to prenatal testing. The numbers refer to the approximate percentages of women taking these options. Termination of pregnancy (TOP) is possible at any of these stages within the limitations imposed by State and Territory laws.

*Note that the timing for offering a CVS in South Australia is sometimes from 10 weeks gestation.*
### Table 1. Advantages and disadvantages of screening tests during pregnancy

<table>
<thead>
<tr>
<th>Screening test *</th>
<th>Gestation (weeks)</th>
<th>% Down syndrome pregnancies detected</th>
<th>Advantages</th>
<th>Disadvantages</th>
</tr>
</thead>
</table>
| Combined first trimester screening | 10 - 12 (blood test) 11³ - 13⁶ (ultrasound with nuchal translucency) | 85 - 90% | • Early screen and therefore early diagnosis  
• Highest detection rate  
• No added risk of miscarriage  
• Detection of some fetal abnormalities  
• Benefits relating to early scan:  
  > Accurate dating  
  > Diagnosis of multiple pregnancy  
  > Diagnosis of early pregnancy failure (miscarriage) | • Will detect some affected pregnancies that may spontaneously miscarry  
• Does not provide risk for neural tube defects but the ultrasound may detect anencephaly  
• Women may not access services so early in the pregnancy  
• Ultrasound requires accredited operator for accuracy  
• Out-of-pocket expenses vary |
| Second trimester maternal serum screening | 14 - 20 (15 - 17 ideal) | 70 - 75% | • Available to women presenting in second trimester  
• No added risk of miscarriage  
• No out-of-pocket expenses for public patients if arranged through public hospital | • Later screening test  
• Inaccurate dates can result in inaccurate risk by calculations. A dating scan should be considered if dates are uncertain  
• Lower detection rate  
• No neural tube risk can be given if test done at 14 weeks  
• Out-of-pocket expenses vary |

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*a* False positive rate set at 5% at these detection rates  
*b* Nuchal translucency measurement should be performed by a Fetal Medicine Foundation (FMF) and RANZCOG (Royal Australian and New Zealand College of Obstetrics and Gynaecology) accredited operator  
*c* May be limited access in some states: in Victoria, it is currently not funded for public patients; in Queensland the blood test is not available publicly  
*d* Assumes the use of the quadruple test (four analytes) and ultrasound dating
Table 2. Advantages and disadvantages of diagnostic tests during pregnancy

<table>
<thead>
<tr>
<th>Diagnostic Test</th>
<th>Gestation (weeks)</th>
<th>% Down syndrome pregnancies detected</th>
<th>Advantages</th>
<th>Disadvantages</th>
</tr>
</thead>
</table>
| Chorionic villus sampling (CVS) * | From 11 weeks b | > 99% | • Early detection  
• Definitive diagnosis  
• Results potentially available in time for termination of pregnancy (TOP) by curette | • Miscarriage risk (~ 1% above background in expert hands)  
• Detects chromosomally abnormal pregnancies that may otherwise spontaneously miscarry  
• 1% risk of equivocal results (placental mosaicism or maternal cell contamination of sample)  
• 0.1% failure to detect chromosome abnormality (abnormality is present in fetus but not in placenta, or maternal cell contamination of sample) |
| Amniocentesis a | From 15 weeks c | 100% | • Test with lowest miscarriage rate  
• Definitive diagnosis | • Miscarriage risk (~ 0.5% above background in expert hands)  
• Diagnosis in second trimester, when pregnancy is more established  
• Results available too late for TOP by curette – TOP may need to be performed by induction of labour or vaginal evacuation c |
| Second trimester fetal anomaly ultrasound scan*  
(*also considered a screening test, see Disadvantages) | 18 - 20 | Very low pick up rate on soft markers alone | • Detects many physical fetal abnormalities, such as: neural tube, cardiac, limb, gastrointestinal, CNS  
• No added risk of miscarriage  
• Measures fetal growth and locates position of placenta | • Not all physical abnormalities can be detected  
• ‘Soft markers’ (risk factors for chromosomal abnormalities not definitive and difficult to interpret) – 50% of babies with Down syndrome will have soft markers/signs  
• Not recommended as primary screening test for Down syndrome |

a Out-of pocket expenses vary for CVS, amniocentesis and scans according to state. Public patients attending a tertiary public hospital may not be charged if identified as ‘increased risk’
b The timing of CVS is not uniform throughout Australia, eg in South Australia it is sometimes offered from 10 weeks gestation  
c Procedures after 20 weeks gestation may not provide results in timeframe permitting second trimester termination of pregnancy. Refer to your State abortion laws, and policies of local perinatal units
**Offering testing**

- All pregnant women or couples contemplating pregnancy should be offered information regarding screening tests. Women should be informed that they will be offered further testing if they have an increased risk result on a screening test.
- All women at increased risk should also be offered information about diagnostic tests.
- All women/couples undertaking screening and diagnostic tests should be made aware that there could be an unanticipated finding. For example, testing for Down syndrome may identify a fetus with Turner syndrome.
- If a woman decides to have prenatal testing, the best test will depend on the woman’s gestation, risk and her concerns. Access to services may also influence the decision. The decision about which test is best is a personal one for each woman/couple.
- Not all tests are available in the public sector and some tests do not have a Medicare rebate.
- Risk can be difficult for individuals to understand (see ‘Ways of explaining prenatal risk figures’ below).

**Prenatal screening tests**

- Types of prenatal screening tests include:
  - Ultrasound scanning
  - Combined first trimester screening
  - Second trimester maternal serum screening

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**Important considerations about prenatal screening tests**

- Screening tests can determine who is at increased risk of having a baby with Down syndrome. Women choosing screening tests should be informed that they will be offered further testing if they have an increased risk and that they may choose to proceed to diagnostic testing. Reasons for not having diagnostic testing include concern about the risk of miscarriage, not wishing to know prior to the birth, and termination of pregnancy being unacceptable to the family.
- Screening tests are non-invasive so there is no increased risk of miscarriage from the procedure.
- Every screening test has a ‘false positive’ rate, where women receive an increased risk result even though their baby is unaffected. This rate is usually around 5%.
- In the majority of pregnancies with an increased risk result on a screening test, the baby is unaffected. A common misconception held by women is that screening tests ‘show’ that the fetus has Down syndrome. Anxiety at any increased risk result is normal (see, *Genetics in practice*).
- Low risk results do not exclude Down syndrome or other abnormalities.
- A second trimester ultrasound may detect some birth defects but is not recommended as a screening test for Down syndrome.
- Neural tube defects and some other conditions may also be detected with second trimester maternal serum screening.
Ultrasound scanning

- Ultrasound scans are screening tests for some birth defects, with varying degrees of accuracy depending on the condition.
- Ultrasound scans can be a diagnostic tool for some birth defects (e.g., neural tube defects).
- Ultrasound scans are non-invasive, and current evidence supports that they pose no threat to the baby.
- Ultrasound screening for fetal anomaly should not be considered a routine test, but should be offered as a choice for all pregnant women.

First trimester ultrasound

- Usually between 8 and 11 weeks gestation.
- Used to:
  > Confirm gestational age
  > Check the pregnancy when there has been a complication such as bleeding
  > View the position of the placenta
  > Confirm presence of multiple pregnancy
  > Check fetal growth, physical development and viability

Figure 3. First trimester ultrasound

Second trimester ultrasound

- Usually at 18 to 20 weeks gestation.
- This ultrasound for fetal anomaly should not be considered a routine test, but should be offered as a choice for all pregnant women.
- May be used to detect:
  > Neural tube defects (anencephaly, spina bifida)
  > Cardiac defects
  > Gastrointestinal malformations (gastrochisis, exomphalos)
  > Limb defects
  > Central nervous system defects
  > Urinary tract anomalies
  > Soft signs that may be associated with underlying chromosomal or other genetic conditions
Limitations of second trimester ultrasound

- Not all malformations can be detected by ultrasound at a second trimester fetal anomaly scan (18 to 20 weeks). The sensitivity depends on many factors including the:
  > Nature of the malformation
  > Experience of the operator
  > Position of the placenta
  > Maternal weight (obesity)
  > Resolution of the ultrasound equipment

- Visualisation of the fetus can be affected by factors such as the woman’s build and the position and size of the fetus.

- Second trimester ultrasound scan is not recommended as a primary screening test for Down syndrome.

- The significance of changes (soft signs of chromosome abnormality) detected by ultrasound may be difficult to interpret. Further investigations may be indicated. Advice can be sought from a specialist ultrasonographer, Genetics Services or an obstetrician in a high-risk pregnancy management unit.

Ultrasound and neural tube defects

- The best detection method for neural tube defects is an ultrasound scan during the second trimester (18 to 20 weeks).

- If a previous pregnancy has been affected by a neural tube defect or there is a 1° relative with a neural tube defect, a specialised ultrasound scan at both 12 to 13 weeks for anencephaly and 18 weeks for other neural tube defects, is recommended.

- The identification of a neural tube defect by ultrasound depends on the skill of the operator, the equipment, the position and gestation of the fetus, and maternal conditions.
Nuchal translucency screening

- Nuchal translucency (NT) describes the appearance of a fluid-filled space at the back of the fetal neck that can be seen using ultrasound early in pregnancy. The depth of the fluid in this space can be measured using ultrasound. The thicker the nuchal translucency the greater the risk of fetal anomalies such as Down syndrome, other chromosomal conditions, cardiac defects and some rare genetic conditions (see *Chromosomal conditions*).
- NT measurements should only be performed by trained and accredited operators, using a risk assessment program that incorporates NT, Crown-Rump Length (CRL) and maternal age. This test should be done when the fetus has a CRL of 45 to 84mm, which corresponds to the period 11 weeks 3 days to 13 weeks 6 days.

It should be noted that nuchal translucency screening alone is not recommended as a screening test for Down syndrome and that it should be combined with a biochemical test (see below ‘Combined first trimester screening’) if available.

Combined first trimester screening

- It is recommended that the NT screening test be done in conjunction with a maternal blood test. Two proteins present in the maternal blood are measured. These are PAPP-A (pregnancy associated plasma protein) and free β-subunit of human chorionic gonadotrophin (free β-hCG). Levels of these proteins vary, but tend to be different in women who are carrying fetuses with Down syndrome or trisomy 18. Increased free β-hCG with decreased PAPP-A is suggestive of Down syndrome, while decreased levels of both analytes is suggestive of trisomy 18.
- By having the blood test in combination with the NT screening test, around 85-90% of babies who have Down syndrome and occasionally other problems will be picked up, compared to 70% or less using NT on its own.
- Approximately 5% of combined first trimester screening tests give an increased risk result. This figure varies depending on maternal age. Women with an increased risk result should be offered a diagnostic test. The majority of increased risk results are not due to Down syndrome, and most of these babies will be healthy.
- Results are usually available on the day of the NT ultrasound, if blood was collected prior to the ultrasound at 10 to 12 weeks gestation, although this will depend on the local provider.
- Results are provided as risks for Down syndrome and the other chromosomal trisomy 18, *at the time of screening* 1. Note then that this is not a risk of delivering an affected fetus. Approximately 30% of babies with Down syndrome do not survive to term.
- Depending on the State/Territory, combined first trimester screening is not always available in the public sector, and there may be out-of-pocket costs for the patient (refer to Table 1).

1 However, in Western Australia the risk figure is adjusted to give the risk at the time of delivery.
Arranging combined first trimester screening

- Tests should be arranged a couple of weeks in advance to allow time to coordinate the blood test and ultrasound. The blood test should ideally be performed first.
- Arrange the ultrasound scan with a Fetal Medicine Foundation (FMF) accredited operator (see Table 1).
- Arrange the blood collection at an appropriate gestation (see Table 1). Blood can be collected at the local pathology service but the request should have clear instructions for the sample to go to the screening lab. This process varies between States, private and public sectors, and metropolitan and regional centres. For specific details regarding co-ordinating the results of the blood and nuchal translucency test contact your local FMF accredited operator of choice.

The factors that need to be entered into the risk calculation algorithm should be noted on the request form including:

- LMP & EDD
- Current weight
- Maternal age
- Previous child with a chromosomal abnormality
- Date and location of ultrasound scan
- Any other information requested on the form, eg ethnicity, IVF details

a If the results of the scan are not received by the date on the form, the laboratory will contact the ultrasound practice or the requesting doctor.

Second trimester maternal serum screening

- The optimal time to have this test performed is between 15 and 17 weeks, but it can be performed until 20 weeks.
- Second trimester maternal serum screening uses a blood test in conjunction with the maternal age, gestational age and maternal weight to calculate a risk figure for Down syndrome. This screening test may also detect pregnancies with an increased risk for trisomy 18 (see Chromosomal conditions) and neural tube defects.
- Maternal blood contains hormones and proteins produced by the fetus and placenta, including alpha-fetoprotein (AFP), unconjugated estriol (μE3), free β-subunit of human chorionic gonadotrophin (free β-hCG), and inhibin A.
- Levels tend to be altered in pregnancies affected by Down syndrome, trisomy 18 or neural tube defects. In Down syndrome, the levels of AFP and μE3 tend to be reduced, and free β-hCG and inhibin A increased. In neural tube defects AFP may be increased and, in trisomy 18, levels of all these substances are decreased.
- The number and type of analytes used may vary between pathology services. The quadruple test measures four analytes (AFP, μE3, free β-hCG and inhibin A), whilst the triple test measures three analytes. Detection rates are improved when four analytes are used.
- Using the quadruple test with ultrasound dating:
  > 75% of fetuses with Down syndrome are detected by second trimester maternal serum screening, and approximately 5% of tests give an increased risk result. Women aged 40 and over have higher detection rates, with 95% of affected pregnancies receiving an increased risk result. At least 50% of all women in this age group will receive an increased risk result.
  > 85% of babies with a neural tube defect will be detected using maternal serum screening alone. When combined with a detailed ultrasound, the detection rate for spina bifida can be as high as 95%, and 100% for anencephaly.
- Results are usually available within 24 to 48 hours of collection, but may take longer in parts of regional Australia. Women at increased risk are offered diagnostic testing. The majority of increased risk results are not due to Down syndrome and most of these babies are unaffected. The possibility of false positive results and the management options should be discussed with women prior to screening, as should the fact that this blood test cannot definitively identify babies with Down syndrome, trisomy 18, or neural tube defects.
Arranging second trimester maternal serum screening

The factors that need to be entered into the risk calculation algorithm should be noted on the request form:

- Ultrasound-based gestation or LMP
- Current weight
- Maternal age
- Previous child with a chromosomal abnormality as well as previous child or close relative with a neural tube defect
- Date of collection
- If the woman has insulin-dependent diabetes
- And any other information requested on the form, eg ethnicity, IVF details

- Some States’ and Territories’ Antenatal Screening Programs receive government funding to perform this testing on public patients, and so there is no out-of-pocket cost.
- For private patients, the cost depends on the pathology provider.
- This test does not need to be repeated unless blood was collected at less than 14 weeks gestation.

Counselling for an increased risk result

- Cut-off risks are chosen to decide who is at an increased risk as a result of screening. Women at increased risk can then be offered diagnostic testing. Cut-off risks for trisomy 18 and Down syndrome are listed below. For neural tube defects an AFP level is measured in MoM (multiple of the population median, corrected for gestation). Note that there is some State variation.

Table 3. Cut-off risks used to determine increased risk in prenatal screening

<table>
<thead>
<tr>
<th>Screen</th>
<th>Down Syndrome</th>
<th>Trisomy 18</th>
<th>Neural tube defect</th>
</tr>
</thead>
<tbody>
<tr>
<td>Combined first trimester screening</td>
<td>1 in 300</td>
<td>1 in 300</td>
<td></td>
</tr>
<tr>
<td>Second trimester maternal serum screening</td>
<td>1 in 300 a</td>
<td>1 in 300</td>
<td>≥ 2MoM c</td>
</tr>
</tbody>
</table>

a Cut-off in Victoria is 1 in 250
b Cut-off in Victoria is 1 in 200, cut-off in NSW is 1 in 250
c Cut-off in Victoria is ≥ 2.5MoM

- First confirm the gestational age as incorrect gestational age estimation can affect accuracy of results.
- It may be helpful to discuss the results with the service provider prior to informing the woman/couple.
- Listen and give the woman/couple time to absorb the news, consider available options and to make decisions about further testing. Informed decision-making is assisted by the provision of up-to-date, unbiased information.
- Ensure the woman understands that while the pregnancy is at increased risk for the condition, the result is not definitive.
- A common misconception held by women is that screening tests ‘show’ the fetus has Down syndrome. It can take some listening, clarification and explanation to counteract this belief. Anxiety at any increased risk result is normal (see Genetics in practice).
- Reassure the woman that the majority of babies with an increased risk result will be normal and healthy.
- Discuss the option of diagnostic testing. Not all women decide to proceed to diagnostic testing. Reasons for not having diagnostic testing include concern about the risk of miscarriage, not wishing to know prior to the birth, and termination of pregnancy being unacceptable.
  > Post-test counselling is available from specialist obstetricians and Genetics Services. Referral should be considered for discussion of diagnostic tests and counselling for women with increased anxiety.
Ways of explaining prenatal screening risk figures

Giving an increased risk result:
A 29 year old woman receives an increased risk result for Down syndrome after a serum screen test. The risk is 1 in 100. Prior to the test her risk was based on maternal age alone and was 1 in 1002.

- Comparison to other women getting the same test result:
  “Of 100 women with this test result, on average, one will have a baby with Down syndrome and 99 will have babies who do not have Down syndrome.”

- Risk relative to maternal age:
  “Your risk is now similar to that of a 39 to 40 year old who has not had any screening. Women who have this level of risk are offered further testing to determine if the baby has Down syndrome.”

Prenatal diagnostic tests

- Types of prenatal diagnostic tests include:
  > Chorionic villus sampling (CVS)
  > Amniocentesis
  > Ultrasound

Important considerations about prenatal diagnostic tests

- CVS and amniocentesis testing require invasive sampling procedures to obtain cells for chromosome analysis or specific genetic tests.
- It is not possible to detect or diagnose every possible condition a child might have using prenatal diagnosis.
- Both CVS and amniocentesis have a risk of miscarriage. The risk is operator dependent, so tests should be performed by an experienced operator.
- Indications for offering diagnostic testing include:
  > Advanced maternal age (≥37 yr in Victoria, ≥35 yr elsewhere)
  > A previous pregnancy with a chromosome abnormality
  > The presence of soft signs of chromosome abnormality during ultrasound examination
  > A parent carrying a chromosome translocation (5% of Down syndrome is caused by an unbalanced translocation involving chromosome 21 that was inherited from a parent with a balanced form of the translocation, see Chromosomal conditions)
  > An increased risk result on a screening test
  > An increased risk of having a baby with a genetic condition – usually when there is a family history of an inherited genetic condition
- Prior to testing, there should be a full discussion of the advantages and disadvantages of the procedures, the implications of the possible test results and subsequent management options, including methods of termination of pregnancy and the available supports.
Chorionic villus sampling (CVS)

- This procedure is usually performed from 11 weeks, routinely between 11 and 13 weeks gestation. It should not be performed prior to 10 weeks gestation due to the risk of limb defects.
- A sample of chorionic villus (pre-placental tissue) is removed by a fine needle, either transabdominally, or less frequently transvaginally, under ultrasound guidance (see Figure 6).
- The tissue is used for chromosome analysis, and in some specific situations, may be used for diagnosis (DNA or biochemical analysis) of a genetic condition where there is a family history.
- It is a procedure that can be performed earlier in pregnancy than amniocentesis, and has the benefits of an early scan that may detect anencephaly.
- Some women experience cramping and, occasionally, vaginal bleeding after CVS.
- The miscarriage rate, in experienced hands, is estimated at ~1% above the background risk.

In South Australia CVS is sometimes performed between 10 and 11 weeks

Important points about CVS

- CVS has a 1% risk of equivocal results. This includes the risk of placental mosaicism – the presence of a mixture of cells with normal and abnormal karyotype, or maternal cell contamination of the sample. In this case, amniocentesis may be necessary to clarify the karyotype of the fetus.
- CVS has a 0.1% rate of failure to detect a pregnancy with a chromosomal anomaly, due to the occasions when there is an abnormal karyotype in the baby, but not in the placenta.
- CVS may not detect fetal chromosomal mosaicism.
- Test results take about one week.

Figure 6. CVS procedure
Amniocentesis

- The procedure is performed from 15 weeks routinely, to approximately 19 to 20 week gestation.
- A sample of amniotic fluid is removed from around the fetus by a fine needle, under ultrasound guidance (see Figure 7).
- Testing is performed on amniotic fluid cells, most of which originate from the fetus, compared with CVS, where placental cells are tested.
- Test results take between one and three weeks.
- Discomfort is usually minimal, though a very small number of women experience pain as the needle passes through the peritoneum.
- The miscarriage rate is lower than that for CVS and estimated to be around 0.5% above the background risk in experienced hands.
- Amniocentesis may not detect mosaicism, because of the limited number of cells counted in a routine test.
- As amniocentesis is a second trimester test, the pregnancy is more advanced when results become available, compared with CVS.
- NB: amniocentesis is not the preferred screening test for neural tube defects. An ultrasound at 18 to 20 weeks gestation is more accurate.

Arranging CVS and amniocentesis

- Refer to a private ultrasound practice or public hospital ultrasound department.
- The woman’s blood group should be given on the request form, as rhesus negative women will require an anti-D injection.
- There are no out-of-pocket costs in the public system if there is an indication for testing.
- For private patients, there are costs for both the sampling procedure and chromosome analysis. Contact service providers for details listed at the end of this section.
Ultrasound diagnostic testing

- May be used to detect:
  - Neural tube defects (anencephaly, spina bifida)
  - Cardiac defects
  - Gastrointestinal malformations (gastrochisis, exomphalos)
  - Limb defects
  - Central nervous system defects
  - Urinary tract anomalies
  - Soft signs that may be associated with underlying chromosomal or other genetic conditions

Genetic testing

Chromosome analysis

- Prenatal diagnostic testing involves fetal or placental cells being examined to look at the number and structure of each chromosome. A full chromosome analysis, called a karyotype, allows the diagnosis of chromosomal abnormalities. A karyotype takes 7 to 14 days and the result will be sent to the referring clinician.
- Where there are strong indications of a fetal anomaly (eg a markedly increased risk screening result), or where a preliminary result is required quickly, FISH (fluorescent in situ hybridisation) analysis may also be performed on samples obtained using CVS or amniocentesis. FISH results may be available in 1 to 2 days.

Indications for FISH:

- Fetal anomaly detected on routine second trimester ultrasound scan.
- Markedly increased risk result on a screening test.
- Late gestation.
- Parental anxiety.

Benefits of FISH:

- Provides a quick preliminary result for the presence of Down syndrome and certain other chromosome trisomies within 24 to 72 hours.
- FISH will detect chromosome abnormalities. The number of probes used will determine the type of chromosome abnormalities detected.
  - ‘Three-probe FISH’ will detect Down syndrome and sex chromosome abnormalities
  - ‘Five-probe FISH’ will detect Down syndrome, trisomy 18, trisomy 13 and sex chromosome abnormalities

The limitations of FISH

- FISH can give false positive results, and thus an abnormal result needs to be interpreted cautiously if other indications of trisomy are not present.
- FISH can give false negative results. A normal FISH result only excludes full trisomies of the chromosomes tested (usually 13, 18, 21, X, Y). FISH usually cannot rule out structural abnormalities of these chromosomes, nor trisomies of other chromosomes.
- FISH does not replace a complete chromosome analysis (karyotype) and this should still be completed.
- FISH results may be inconclusive if both normal and abnormal cells are present (mosaicism).
Arranging a FISH test
- FISH is requested by the obstetrician or the ultrasonographer.
- There is no Medicare rebate for FISH.
- The charge for FISH depends on the laboratory doing the testing and the number of probes used.
- Costs for three-probe FISH may be less than for five-probe FISH.
- Contact the pathology or ultrasound service for details of costs.
- Results are usually available in 24 to 72 hours.

Genetic conditions requiring DNA testing
- A woman/couple with a family history of a genetic condition must be referred to Genetics Services as soon as possible, preferably prior to pregnancy.
- Testing for inherited conditions (eg cystic fibrosis or thalassaemia) requires knowledge of the causative genetic mutation in the family. This may require extensive, time-consuming tests before a prenatal test can be offered. Referral once the woman is pregnant may be too late to offer prenatal diagnosis.
- In cases of rarer conditions the laboratory may need to be notified in advance. Some tests are only available interstate or even overseas, and much co-ordination is needed for shipping and testing to ensure a timely result.
- CVS is the preferred diagnostic procedure when a pregnancy is known to be at increased risk of a genetic condition. Prenatal diagnosis of genetic conditions requires the coordination of the sampling procedure, cytogenetic laboratory and DNA laboratory. The woman/couple should therefore be referred to Genetics Services as soon as the pregnancy is confirmed. The time until results are available will depend upon the type of test performed.

Pre-implantation genetic diagnosis (PGD)
- PGD is the genetic testing of embryos prior to implantation in the womb and relies on usual IVF techniques to generate embryos in vitro. Embryos are usually tested at day 3 (6 - 10 cells) after fertilisation. If fertilisation using conventional IVF is unsuccessful, ICSI (Intra Cytoplasmic Sperm Injection) technology may be used.
  > As shown below, pre-implantation genetic diagnosis uses assisted reproduction technology (ART). Hormones are used to stimulate a woman’s ovaries and enable the collection of a number of eggs or oocytes. After the eggs are removed, the eggs are fertilised in the laboratory with sperm. Those eggs that are successfully fertilised are allowed to divide and multiply for 3-6 days, by which stage they contain about 8 cells or have developed into a blastocyst. Not all fertilised eggs make it to this stage.
  > One or two cells are removed in order to test for the specific genetic condition in question. The removal of these cells does not appear to harm the developing embryo. Only those embryos that do not appear to be affected will be transplanted into the mother’s uterus usually on the same day. Generally, no more than one or two embryos will be transferred to the uterus at any one time to avoid the possibility of multiple births. In some IVF centres, unaffected embryos that are not used can be frozen for transfer in another cycle.

The PGD process
Tests that can be performed
- Identification of sex for diagnostic reasons but not for family balancing.
- FISH analysis to identify chromosome trisomies and translocations.
- DNA testing for selected single gene conditions.
- Testing can only be performed if the gene mutation(s) causing the condition are known and testing for the mutation(s) is accurate on a single cell.
Advantages of PGD

- If a woman and her partner are trying to avoid a pregnancy affected with a certain genetic condition, the risk of this can be minimised without termination of the pregnancy.
- Referral for specialised counselling is required. This provides a forum to discuss issues in detail with an experienced counsellor who has been trained in this area.

Limitations of PGD

- There is no guarantee of achieving a pregnancy. Most couples undergo several cycles of treatment before achieving an ongoing pregnancy.
- Accuracy is high, but not 100%.
- The woman must undergo IVF procedures.
- The procedures and testing are expensive.
- It is time consuming and requires meticulous lab work.
- There is a risk of multiple pregnancy if more than one embryo is implanted.

Counselling issues

- Risks and success rates of PGD must be made clear in relation to other methods of avoiding genetic risk.
- There can be personal moral dilemmas regarding the use of embryos not implanted.
- Grief and loss.
- Attitude to prenatal diagnosis
Managing a pregnancy with an abnormal karyotype

(See also Genetics in practice.)

- It may be helpful to discuss the results with the service provider or clinical geneticist prior to informing the woman/couple.
- Pre-test counselling should prepare the woman/couple for the possibility of a chromosome abnormality which may not have been anticipated.
- Listen and give the woman/couple time to absorb the news and consider their options.
- Referral to specialist Genetics Services is recommended for sex chromosome abnormalities and mosaic results and should be considered for all other abnormal karyotypes.
- Not all chromosome abnormalities have a major effect on the baby. Medical texts are often out of date. It is important that the woman/couple receives up-to-date, unbiased information about the potential effects of a chromosome abnormality. Discussion with support groups can be helpful for parents (see Contacts, support and testing).
- Decisions regarding the pregnancy should only be made once there has been a full discussion of the implications of the test results and the management options. This might include referral to Genetics Services, discussion with obstetricians and paediatricians, and referral to the relevant support group (see Contacts, support and testing).

Managing a pregnancy with a fetal anomaly

- Referral of public patients to a high-risk clinic, perinatal management unit or fetal diagnostic unit is strongly recommended for a management plan and coordination of ongoing care. Further prenatal testing may be needed to clarify the diagnosis, eg abdominal wall defect may be due to a chromosomal trisomy.
- Pre-test counselling should prepare the woman/couple for the possibility of a structural abnormality which may be detected incidentally.
- Women and families can benefit from detailed discussion with support groups. The Fetal Medicine or Genetics Services will usually arrange for the family to meet clinicians who have experience in the management of babies with disability and birth defects. These services are located in the public and private sector.
- A management plan may include providing the couple with:
  - The opportunity for consultation with appropriate specialists
  - Further diagnostic procedures
  - Further ultrasound scans in the presence of the appropriate clinical specialist (eg cardiologist present if cardiac defect is suspected)
  - Genetic counselling
  - Ongoing support if the pregnancy continues to term
  - Specific plans for delivery, postnatal care and support
  - Contact details of the relevant support groups
Counselling regarding termination of pregnancy

- It is common for women/couples to be deeply shocked after receiving difficult/bad news. They often immediately request a termination. It is important to allow some time for the woman/couple to come to terms with the news and consider their decision. This may include discussions with relevant health professionals and support groups.
- If termination of pregnancy is an option, the woman/couple should be encouraged to make their decision based on their personal values and accurate, up-to-date and unbiased information.
- When women/couples have a choice of the method of termination, all options and the associated risks and advantages should be discussed, preferably with an obstetrician or prenatal Genetics Services.
- Whether or not they wish to see the baby after termination should be discussed in advance.
- If a post-mortem examination is required for accurate diagnosis, this should also be discussed in advance.
- Grief after a termination is a normal reaction to the loss of a wanted pregnancy and can be complicated by feelings of guilt and anxiety for future pregnancies.
- Ongoing support for the woman/couple is important, regardless of their decision. Women/couples may turn to their GP for support, or benefit from consultation with a genetic counsellor with experience in prenatal diagnosis. Contact your local AGA Peak Body (see Contacts, support and testing) for a local support group.

Management of a pregnancy identified as increased risk by screening, but found to have a normal fetal karyotype

- The woman/couple will experience a raised level of anxiety, even with the best counselling support.
- After a diagnostic test (such as CVS or amniocentesis followed by karyotyping) excludes a chromosomal condition (Down syndrome, trisomy 18 or 13), the pregnancy remains at increased risk of other fetal anomalies or obstetric complications.
- All women with increased risk following screening tests, but normal fetal karyotypes, should be referred for an 18 to 20 week detailed morphology ultrasound, and be monitored closely throughout the remainder of the pregnancy.
**Frequently asked questions**

*What if the woman presents late?*
A woman who is not at increased or high risk of having a baby with Down syndrome and first presents after 20 weeks gestation is limited to an ultrasound scan for fetal anomalies, which should be immediately arranged.

For a woman aged 35 years and over (or 37 in Victoria) at EDD, an amniocentesis with FISH could be considered up to the end of the 20th week, although terminations of pregnancy are usually required to be done at less than 20–22 weeks. This may vary in different States and Territories according to their laws.

FISH analysis could be performed to give a preliminary result within 48 hours of the amniocentesis, with the final karyotype result taking up to two weeks. The pregnancy is then fairly advanced and termination of pregnancy may not be available as an option.

*What do I say to a woman who is anxious while waiting for her results?*
From the time the test is arranged, it is important that the woman be given accurate information and realistic expectations, including the maximum period to wait for test results. She should be informed that the time taken to receive results does not indicate if they will be normal or not. Talking about the most likely outcomes can be helpful, but avoid false reassurance. Often listening to the woman’s anxieties and using counselling skills such as active listening, normalising and acknowledging the distress and uncertainty are useful. Make information available upon request.

*What do I say to a woman whose Down syndrome risk has increased, but is still below the cut-off for a diagnostic procedure?*
Explore the meaning of this result for the woman. While her risk is greater than other women her age, her risk of having a baby with Down syndrome is still low. Some women may not be prepared to accept a certain level of risk and may still choose to have a diagnostic test.

*Can a woman have both first and second trimester screening tests?*
Intuitively, more screening will identify abnormalities better – however this does not occur in practice. For a marginal increased detection rate for Down syndrome, the false positive rate will rise substantially. If a woman has both screening tests, she is more likely to be identified at increased risk, and offered an invasive diagnostic procedure. The more invasive procedures performed, the greater the risk of causing spontaneous loss of an unaffected fetus. Therefore, it is strongly recommended **not** to have both first and second trimester screening tests.
Correcting misunderstandings

‘I’m young, so I don’t need to have any tests for Down syndrome’
Although younger women are at lower risk than older women, many babies with Down syndrome are still conceived by women who would not be defined as at increased risk due to their age (ie 35 or 37 years and above). Screening tests should be offered to women of all ages.
Diagnostic testing can follow if requested.

‘The tests were all OK, so my baby is normal’
Tests during pregnancy can detect increased risk or presence of certain conditions only. No test, or combination of tests, will detect all birth defects or medical conditions.

‘My blood test was normal, so my baby doesn’t have Down syndrome’
Blood/screening tests cannot detect all pregnancies with Down syndrome. A woman with a ‘normal’ (low risk) screening test result does have a chance of having a baby with Down syndrome but this risk is not high enough for diagnostic testing to be indicated.

‘The blood test says there’s something wrong with my baby’
Blood/screening tests during pregnancy do not detect birth defects; they indicate which pregnancies have an increased risk of certain genetic conditions and birth defects. Most fetuses with ‘abnormal’ (increased risk) test results do not have Down syndrome. This is an indication for referral for diagnostic procedures.

‘The blood test says there is something wrong so I need a diagnostic test’
If the blood/screening test was second trimester maternal serum screening, increased risk results are due to inaccurate dates (if LMP only given) in 30% of cases. Check dates by ultrasound. An increased risk result on a screening test is an indication for diagnostic testing; however, a small number of women choose not to have diagnostic testing.

‘If I have another screening test, I might get a better result’
Screening tests are most accurate when done at the correct time in the pregnancy. Retesting is only performed if dates are inaccurate. It is strongly recommended not to have both first and second trimester screening tests (see above, ‘Frequently asked questions’).

‘I am over 35 years so I need to have a CVS or amniocentesis’
Women aged 35 years and over (or 37 years in Victoria) at EDD may choose to have a diagnostic test, may prefer the option of screening tests, or may choose to have no tests at all. Women in this age group should be aware that they are more likely to have an increased risk result from a screening test as age is part of the risk calculation, in which case they will then need to consider diagnostic testing.

‘It’s not worth having any tests because I wouldn’t terminate the pregnancy’
Some people feel it is beneficial to know if the fetus has an anomaly to prepare for the birth and future. Others prefer to wait until delivery. All women should have the opportunity to consider testing.
List of fetal medicine services in public hospitals associated with the state genetics services in Australia

### Australian Capital Territory

<table>
<thead>
<tr>
<th>Hospital</th>
<th>Address</th>
<th>Contact Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>The Canberra Hospital</td>
<td>Genetic Counsellor, PO Box 11, Woden ACT 2605</td>
<td>Ph: (02) 6244 2133 Fax: (02) 6244 4625</td>
</tr>
</tbody>
</table>

### New South Wales

<table>
<thead>
<tr>
<th>Location</th>
<th>Hospital</th>
<th>Address</th>
<th>Contact Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>Camperdown</td>
<td>Royal Prince Alfred Hospital</td>
<td>Department of Molecular and Clinical Genetics, Building 65, Level 6 Missenden Road, Camperdown NSW 2050</td>
<td>Ph: (02) 9515 5080 Fax: (02) 9550 5389</td>
</tr>
<tr>
<td>Kogarah</td>
<td>St George Hospital</td>
<td>Women and Children’s Health Gray Street, Kogarah NSW 2217</td>
<td>Ph: (02) 9113 3635 Fax: (02) 9113 3694</td>
</tr>
<tr>
<td>Liverpool</td>
<td>Liverpool Hospital</td>
<td>Fetal Medicine Unit Locked Bag 7103, Liverpool BC NSW 1871</td>
<td>Ph: (02) 9828 5631 Fax: (02) 9828 5570</td>
</tr>
<tr>
<td>Newcastle</td>
<td>John Hunter Hospital</td>
<td>Maternal and Fetal Medicine Locked Bag 1, Hunter Region Mail Centre, Newcastle NSW 2310</td>
<td>Ph: (02) 4921 4694 Fax: (02) 4921 3133</td>
</tr>
<tr>
<td>Penrith</td>
<td>Nepean Hospital</td>
<td>Perinatal Ultrasound Level 3 South Block, Derby Street, Penrith NSW 2751</td>
<td>Ph: (02) 4734 2578 Fax: (02) 4737 3206</td>
</tr>
<tr>
<td>Randwick</td>
<td>Royal Hospital for Women</td>
<td>Maternal/Fetal Medicine Barker Street, Randwick NSW 2031</td>
<td>Ph: (02) 9382 6098 Fax: (02) 9382 6706</td>
</tr>
<tr>
<td>St Leonards</td>
<td>Royal North Shore Hospital</td>
<td>Fetal Medicine Unit Pacific Highway, St Leonards NSW 2065</td>
<td>Ph: (02) 9926 6478 Fax: (02) 9926 7880</td>
</tr>
<tr>
<td>Westmead</td>
<td>The Children’s Hospital</td>
<td>Department of Clinical Genetics Locked Bag 4001, Westmead NSW 2145</td>
<td>Ph: (02) 9845 3273 Fax: (02) 9845 3204</td>
</tr>
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</table>
**Northern Territory**

<table>
<thead>
<tr>
<th>Genetics Service</th>
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<tr>
<td></td>
<td>There is currently no clinical genetics service or outreach service available in the Northern Territory</td>
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</table>

**Queensland**

<table>
<thead>
<tr>
<th>Hospital/Service</th>
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<tbody>
<tr>
<td>Royal Brisbane Women's Hospital</td>
<td>Antenatal Clinic, Cnr Bowen Bridge &amp; Butterfield Rds, Herston Qld 4029 Ph: (07) 3636 2269 Fax: (07) 3636 5379</td>
</tr>
<tr>
<td>Mater Mother's Hospital</td>
<td>Raymond Terrace, South Brisbane Qld 4101 Ph: (07) 3840 1593 Fax: (07) 3840 1621</td>
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**South Australia**

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<thead>
<tr>
<th>Hospital/Service</th>
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<tbody>
<tr>
<td>Women's and Children's Hospital</td>
<td>South Australian Clinical Genetics Service Youth and Women's Health Service, North Adelaide SA 5006 Ph: (08) 8161 7375 Fax: (08) 8161 6088</td>
</tr>
<tr>
<td>Antenatal Diagnosis and Counselling Service</td>
<td>Department of Obstetrics and Gynaecology Women's and Children’s Hospital South Australian Clinical Genetics Service 72 King William Road, North Adelaide SA 5006 Ph: (08) 8161 7633 Fax: (08) 8161 7654</td>
</tr>
<tr>
<td>Perinatal Dysmorphology Group</td>
<td>Department of Obstetrics and Gynaecology Flinders Medical Centre, Bedford Park SA 5042 Ph: (08) 8204 4577 Fax: (08) 8204 3143</td>
</tr>
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**Tasmania**

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<thead>
<tr>
<th>Hospital/Service</th>
<th>Details</th>
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<tbody>
<tr>
<td>Royal Hobart Hospital</td>
<td>Tasmanian Clinical Genetics Service GPO Box 1061L, Hobart Tas 7001 Ph: (03) 6222 8296 Fax: (03) 6222 7961</td>
</tr>
</tbody>
</table>
### Victoria

<table>
<thead>
<tr>
<th>Genetic Health Services Victoria (GHSV)</th>
<th>10th Floor, Royal Children’s Hospital Flemington Road, Parkville Vic 3052 Ph: (03) 8341 6270 Fax: (03) 8341 6390</th>
</tr>
</thead>
<tbody>
<tr>
<td>Monash Medical Centre</td>
<td>Monash Medical Centre 246 Clayton Road, Clayton Vic 3168 Ph: (03) 9594 2026 Fax: (03) 9594 6022</td>
</tr>
<tr>
<td>Royal Women’s Hospital</td>
<td>Specialty Genetics Services 132 Grattan Street, Carlton Vic 3053 Ph: (03) 9344 2121 Fax: (03) 9344 2066</td>
</tr>
</tbody>
</table>

### Western Australia

| King Edward Memorial Hospital for Women | Fetal Medicine Service 374 Bagot Road, Subiaco WA 6008 Ph: (08) 9340 1525 Fax: (08) 9340 1678 |

### Bibliography


### Further information


Scroll down the list to find “Prenatal Testing Special tests for your baby during pregnancy”.
Preparing for pregnancy

Most children born in Australia are born healthy. But about 2 to 3 babies in 100 are born with a condition that means they will need medical care. Some of the conditions can be detected early in pregnancy, while others cannot.

If you are thinking about becoming pregnant, you should talk to your doctor about your particular situation. You may have conditions in the family that you would like to talk about, or you may have health problems, dietary preferences or other issues you wish to discuss.

You should also talk to your doctor about:

- Taking enough of a vitamin called folate, or folic acid. Folate is present in green leafy vegetables, but many women find it hard to eat enough folate naturally. So all women should take extra folate for at least one month before becoming pregnant, as well as for the first three months of pregnancy. This will lower the risk of having a baby with a neural tube defect, which is a problem in the development of the spinal cord (spina bifida) and/or brain (anencephaly).
- Your family history (see fact sheet on ‘Your family history’).
- Your ancestry. For example, certain genetic disorders of the blood cells, known as haemoglobinopathies (see fact sheet 15 on ‘Haemoglobinopathies’), are relatively common in people with a family background from southern Europe, the Middle East, South-East Asia, Africa, the Indian subcontinent, South America, the Caribbean and the Pacific Islands. There are also a number of disorders that are more common in those with Jewish ancestry. People with one of these family backgrounds could be carriers of an altered gene and may wish to consider genetic testing.
- The increased chance for older women of having a baby with Down syndrome or some other chromosome alteration.
- Whether any prescription or other drugs you take could be harmful to a developing baby.
- The potential effects of alcohol and smoking on a developing baby.
- Making sure you are immune to rubella (German measles).

As a result of these conversations, you may benefit from some genetic testing or genetic counselling. Your GP should be able to arrange that, if required.
Contacts and further information

- All states and the ACT have familial cancer services. Contact them through your local state or territory health department.
- Pregnancy and alcohol at http://www.alcoholguidelines.gov.au
- MyDr at http://www.mydr.com.au
- The Centre for Genetics Education at http://www.genetics.edu.au
- HealthInsight at http://www.healthinsite.com
- MedicineNet at http://www.medicinenet.com
- For other related fact sheets, you can contact the Gene Technology Information Service on free call Australia-wide 1800 631 276 or email gsis-australia@unimelb.edu.au or visit Biotechnology Australia’s website at http://www.biotechnology.gov.au
Most children born in Australia are born healthy. But about 2 or 3 in 100 are born with a condition that means they will need medical care.

Some of the more common conditions are:
- Congenital heart disease
- Disorders of the kidney and bladder
- Hip dislocation at birth
- Club foot
- Down syndrome and other chromosome alterations
- Spina bifida and related conditions, which are known as neural tube defects and which are problems in the development of the spinal cord and/or brain
- Cleft lip and/or palate
- Developmental delay.

Some of these can be detected in pregnancy, while others cannot.

Screening tests and/or diagnostic tests are available for some of these disorders. They are not compulsory – it is your choice whether or not to have these tests.

Before having any tests, you need to consider what you would do if a test came back positive or indicates some degree of risk. What would you do if a test showed your unborn child had an abnormality? Would you consider a termination of pregnancy? Would you not consider one? Would you just like to know, even if you don’t plan to do anything about it? Or would you rather not know?

**Screening tests**

These tests do not give a firm diagnosis, but aim to give parents an idea of whether or not they have a higher than normal risk of having a child with the disorder being screened for.

None are perfect – sometimes screening tests miss the conditions they are meant to detect.

If a screening test picks up an increased risk, then there are diagnostic tests – chorionic villus sampling, amniocentesis or ultrasound – that can sort out whether or not the baby has the disorder.
**First trimester screening test**

This test is designed to identify women at increased risk of having a baby with Down syndrome, but it can sometimes also identify other problems.

The test has three parts. The first is a blood test at 10 to 12 weeks of pregnancy. The second is an ultrasound, called a nuchal translucency or NT test, at 11 to 13 weeks. The third part is the woman’s age, which is also taken into account.

These three pieces of information are combined to calculate the risk that the baby has Down syndrome. Couples with an increased risk will be offered genetic counselling to consider their choices; the choice of whether or not to have a diagnostic test – either chorionic villus sampling or amniocentesis – to check the baby’s chromosomes.

**Nuchal translucency test**

Nuchal translucency is used to estimate if a baby is at an increased risk of having a chromosomal abnormality. It uses ultrasound to see and measure a fluid filled sac at the back of the unborn baby’s neck during early pregnancy.

The nuchal translucency test, which is part of the first trimester screening test, can sometimes be done on its own, without the blood test. This ultrasound is carried out between 11 and 13 weeks of pregnancy and is reasonably accurate, but not as accurate as the combined first trimester screening test.

**Second trimester maternal serum screening test**

This blood test is best done between 15 and 17 weeks of pregnancy, but it can be carried out between 14 and 20 weeks. The second trimester screening test is suitable for women who did not have either the first trimester screening test or the nuchal translucency test. It can tell parents whether the baby is at increased risk of Down syndrome (and/or some other chromosomal alterations) or a neural tube defect, which is a problem in the development of the spinal cord and/or brain.

The second trimester test is not as accurate as the first trimester screening test.

**Ultrasound**

Most pregnant women will have an ultrasound at 18 to 20 weeks. This ultrasound checks the baby’s growth, the stage of pregnancy, and the amount of amniotic fluid, the position of the baby and placenta. This ultrasound also looks for physical problems such as neural tube defects, heart and kidney malformations, cleft lip and limb abnormalities.

**Diagnostic tests**

These tests aim to give a firm diagnosis of a potential problem. They are more accurate than screening tests. Like all tests, they are only looking for specific gene alterations or chromosome alterations (see fact sheet on ‘What is a gene?’). They are not perfect and they may occasionally miss something.
**Chorionic villus sampling**

This test can detect chromosomal alterations as well as genetic conditions, which your doctor knows to look for because they have happened before in the family, or because a genetic screening test has shown that you could have an affected baby.

Chorionic villus sampling is usually done at around 11 weeks of pregnancy and preferably by 13 weeks. Usually, a needle is guided through the abdomen to the tissue that will form the placenta, and a small fragment of tissue is removed. Occasionally a plastic tube is guided through the vagina and cervix instead. In both cases, the procedure is monitored by ultrasound so that the needle or plastic tube is kept away from the baby. Most women find it uncomfortable rather than painful.

Women who have chorionic villus sampling have a slightly increased risk of miscarrying afterwards. The risk is between 1 in 100 and 1 in 200.

The results are usually available in two to three weeks.

**Amniocentesis**

This test can detect chromosomal alterations. The test can also detect gene alterations, which your doctor knows to look for because they have happened before in the family, or because a genetic screening test has shown that you could have an affected baby.

Amniocentesis is usually done at 15 to 16 weeks of pregnancy and preferably before 20 weeks. A needle is guided into the fluid around the baby and a small amount of fluid is removed. The procedure is monitored by ultrasound so that the needle is kept away from the baby. Most women find it uncomfortable rather than painful.

Women who have amniocentesis have a slightly increased risk of miscarrying afterwards. The risk is about 1 in 200.

The results are usually available in two to three weeks.

**Ultrasound**

A detailed ultrasound may be used to look for certain disorders that have happened before in the family. Ultrasound can also be carried out at any time if problems arise in the pregnancy.
Contacts and further information

- All states and the ACT have familial cancer services. Contact them through your local state or territory health department.
- Your local hospital antenatal clinic.
- MyDr at http://www.mydr.com.au
- HealthInsite at http://www.healthinsite.com
- For other related fact sheets, you can contact the Gene Technology Information Service on free call Australia-wide 1800 631 276 or email gtis-australia@unimelb.edu.au or visit Biotechnology Australia’s website at http://www.biotechnology.gov.au