



# Prenatal diagnosis

## Screening Tests

Screening tests are non-invasive and indicate the likelihood of having a baby with Down syndrome (this is not a definitive diagnosis), they are often combined and referred to as First Trimester Screening. They include:

- Early Ultrasound or Nuchal Translucency
- Maternal Serum Screening Test (MSS or Triple Test).

South Australian Maternal Serum Antenatal Screening Program [www.wch.sa.gov.au/samsas](http://www.wch.sa.gov.au/samsas)

An increased risk result does not mean the baby will definitely have Down syndrome. Most women in this group are not actually carrying a baby with Down syndrome but will be offered a diagnostic test which is the removal of a sample of cells to confirm.

If the result is 'NOT' at increased risk that means the chance of having a baby with Down syndrome is less than 1 in 300. This does not, however, guarantee the absence of Down syndrome.

## Diagnostic tests

A diagnostic test is the only way to know for sure if you are carrying a baby with Down syndrome.

There are two types of Diagnostic tests:

- Chronic Villus Sampling (CVS) - from 11 weeks
- Amniocentesis - from 15 weeks.

Both diagnostic tests are invasive procedures and involve a needle being inserted into the woman's abdomen so that cell samples can be taken to be examined. The risk of pregnancy loss or miscarriage following these tests are approximately 1 in 200 or 0.5%.

For further information you can have genetic counselling. It is preferable to obtain a referral from your doctor or obstetrician to access genetic counselling.

**Clinical Genetics Services SA**  
Women's and Childrens Hospital  
72 King William Road  
North Adelaide SA 5006  
Ph 8161 7375 or fax 8161 6008

## Outreach clinics are held in

- FMC
- Lyell McEwin Hospital
- Royal Adelaide Hospital

## Outreach regional clinics are held in:

- Mount Gambier
- Port Augusta
- Whyalla

Making decisions about what is best for you involves complex emotional and personal issues. You may find it helpful to discuss these with your partner, family, supportive friends, doctor or obstetrician. Your doctor can refer you to a genetic counsellor who can be helpful both during and after pregnancies. It is preferable to obtain a referral from your doctor or obstetrician to access genetic counselling.

## Continuing a pregnancy

If you have been told pre-natally after a diagnostic test that your baby has Down syndrome, firstly CONGRATULATIONS....We understand that there is a lot to take in so we would encourage you to spend some time looking through our website and the links below.

Remember that Down syndrome is just a small part of who your baby is and will be. No one can predict how much your child will be able to achieve.

The links below offer up-to-date, accurate information about Down syndrome, and about the experiences of living with a son, daughter or sibling with Down syndrome.

## A pregnant mother's guide to Down syndrome

DownSyndromePregnancy.org is a non-profit organization that offers support to expectant parents who have received a prenatal Down syndrome diagnosis and are moving forward with their pregnancy. The material is not appropriate for women who are still weighing options about their pregnancy because it infers that a woman will be continuing a pregnancy to delivery and beyond.

<http://downsyndromepregnancy.org/>

## Brighter Tomorrows

Supporting families with accurate information about Down syndrome.

<http://www.brightertomorrows.org/englishintro.html?language=1>