

## Fluorescent in situ hybridisation (FISH)

This is a special laboratory test that can be performed on both chorionic villus and amniocentesis samples. It is done in addition to the normal cell culture. FISH can rapidly identify approximately 90% of the common chromosome problems (those involving chromosomes 13, 18, 21, X or Y).

3 copies of chromosome 21  
in Down Syndrome >



Coloured "tags" that are specific to each of these chromosomes are added to the fetal or placental cells. Under ultraviolet light the number of "tags" in each cell are counted. In babies with normal chromosomes there should only be two copies of each chromosome (apart from the X and Y). This usually gives a preliminary result in less than 24 hours. The chance of a false FISH result is around 1 in 1000.

## Prenatal Diagnosis Special Tests *for* Your Baby During Pregnancy

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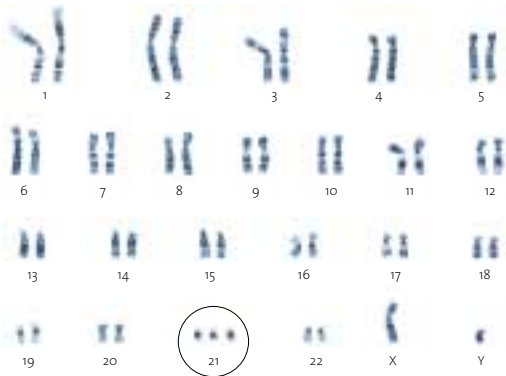
[www.sufw.com.au](http://www.sufw.com.au)

- Chorionic Villus Sampling
  - Amniocentesis
  - Fluorescent in situ hybridisation (FISH)
- 

## Prenatal Testing

Most women enjoy a healthy pregnancy. Some babies, however, are born with problems and about one sixth of these are due to chromosome abnormalities. There are 46 chromosomes in each cell of our bodies, arranged in 23 different pairs with one pair coding for sex (XY for boys and XX for girls). The most common chromosome abnormality is Down syndrome which occurs due to an extra number 21 chromosome.

### Chromosomes in Down Syndrome >



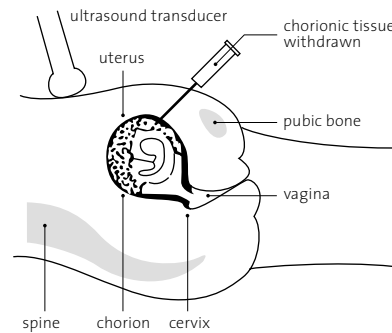
Prenatal diagnosis is available to any pregnant woman considered at increased risk for chromosome abnormality. Risk is increased in women who:

- have an abnormal result on a prenatal screening test (ie fetal nuchal translucency assessment at 11-13 weeks or maternal blood "triple test" at 15-17 weeks)
- are aged 35 or above
- have a family history of a genetic disorder

We can detect chromosome abnormalities and some inherited genetic conditions using chorionic villus sampling (CVS) or amniocentesis.

## Chorionic villus sampling (CVS)

CVS is usually performed between 11-13 weeks of pregnancy.



### How is it performed ?

Using ultrasound guidance, a fine needle passes through the abdominal wall into the placenta and takes a small sample of the tissue (chorionic villi). Sometimes the placenta is difficult to reach through the abdomen and a transvaginal CVS is performed. This procedure is similar to a Pap smear. A fine flexible catheter passes through the cervix into the placental tissue. The discomfort of CVS is usually low.

The baby and placenta come from the same embryonic cells, so they share the same chromosomes. The placental cells are grown in culture to analyse the chromosome number and structure.

### When will the results be available ?

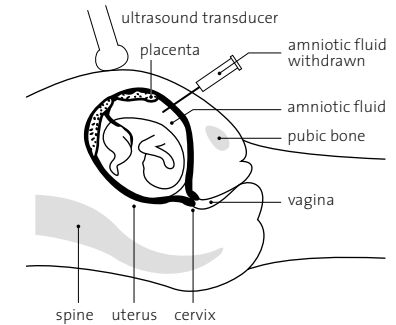
You will be contacted about 9-14 days later with the result. Growing the placental cells in culture takes up most of this time. The test is highly accurate with an error rate of less than 1 in 4000. A preliminary result can be obtained within 24 hours using a new laboratory technique called "FISH" (see over).

### Is there a risk to my pregnancy ?

There is a small risk of miscarriage from the CVS of about 0.5% (1 in 200). This is in addition to the background risk of spontaneous miscarriage happening without testing which is around 2% (1 in 50) at this stage in pregnancy.

## Amniocentesis

This test is able to detect chromosome abnormalities and many cases of spina bifida. It is usually performed between 15-18 weeks of pregnancy.



### How is it performed ?

Using ultrasound guidance, a fine needle passes through the abdominal wall into the amniotic fluid surrounding the baby and a small amount of fluid is removed. There is little discomfort.

The amniotic fluid contains cells from the baby and the lining of the sac. These are grown in culture to analyse the chromosome number and structure. Alpha-fetoprotein (AFP) levels in the fluid are analysed. If the levels are elevated (more than 2.5 times normal) the risk is increased for spina bifida and some other fetal structural abnormalities.

### When will the results be available ?

The AFP result will be available in 2-3 days. In general, you will only be contacted if the level is elevated. The chromosome test will be ready in 11-14 days and you will be contacted with the result as soon as the test is completed. The test accuracy is extremely high with an error rate of around 1 in 12,000. Rapid preliminary assessment with FISH is also available with amniocentesis.

### Is there a risk to my pregnancy ?

Miscarriage may occur following amniocentesis but at SUFW we have been able to minimise this risk. Our own figures show that the risk of miscarriage is as low as 0.25 to 0.33%. That's a 99.75% chance that no miscarriage will occur due to the procedure.