



# Dr. Natalie Shaw

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## Genetic Testing in Pregnancy

There are numerous genetic tests that are available during a pregnancy and even before becoming pregnant.

Some tests can be done on the parents and some test the baby during or after the pregnancy.

### Tests on the unborn baby

Nuchal translucency – this is an ultrasound that is done at about 12 weeks gestation and measures the baby's neck, nasal bone and some hormone levels from the mother. These numbers and the mother's age are calculated together to give a probability of the baby having an extra chromosome 13,18 or 21 (Downs Syndrome). This is not a yes/no answer but is a high quality test. The ultrasound can also see that the baby is developing limbs, heart, kidneys, brain etc. The cost is approximately \$300-\$390 with a medicare rebate of \$60.

Non-invasive Prenatal Test (NIPT, Harmony, Genesyte, Panorama). This is a blood test from the mother where the baby's DNA that is in the mother's blood, is extracted and tested for chromosome 13,18,21 and X and Y. It is 99% accurate if it is normal, however if the result is not normal extra testing may be offered. The blood test is offered by the ultrasound practices such as Ultrasound Care and Sydney Ultrasound for Women for the same cost as the labs charge, but with an ultrasound as well (which is essentially an ultrasound for free). The lab cost is approx. \$450 with no medicare rebate.

CVS/Aminocentesis. These are the invasive tests that are reserved for situations where the NT/NIPT are not normal or there are other genetic diagnoses that need to be made. They involve a needle into the uterus to sample the placenta or fluid for the baby's chromosomes and have a small chance of causing a miscarriage. The test is done at a highly specialised ultrasound practice.

### Tests on the Parents

If there is a family history of genetic illness in your family, you may be offered testing or referral to a geneticist to ascertain which tests may be available (not every illness has a test to check for potential inheritance). These are usually blood tests on the mother or father of the unborn baby.



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Even with no family history of an illness, some people will be silent carriers of genetic illnesses. When someone who is a carrier creates a baby with another silent carrier, there is the potential for the baby to inherit 2 genes that create an illness. An example of this is Cystic Fibrosis, where being a carrier does not affect your health but 2 abnormal genes will lead to the illness in the baby. Tests for some genetic illnesses are available as a blood test from the mother and/or father, such as a newly available test which looks for Cystic Fibrosis/Spinal Muscular Atrophy/Fragile X, costing \$400 with no medicare rebate. There are also more extensive panels of tests available for even less common illnesses, to exclude being a silent carrier of something rarer. Some families may also have had tests done previously or if not, wish to have them arranged, as genetic illnesses can be more common in some communities. An example of this is Tay Sachs disease in the Jewish community. These are again blood tests on the parents.

If both parents are found to be carriers of a genetic illness then testing on the unborn baby such as a CVS or amnio can be arranged to see if the baby is affected by the illness. Some genetic illnesses are so severe that parents consider ending the pregnancy and others may be able to be managed with medication or a special diet such as PKU (Phenylketonuria).

## **Tests of the Newborn Baby**

All newborn babies are offered tests, with their parents' permission, for a range of illnesses which include PKU, Cystic Fibrosis, thyroid disease and other metabolic diseases. This test is a blood test on the baby prior to leaving hospital.