Fact Sheet

Confirmatory Prenatal Diagnosis following Preimplantation Genetic Diagnosis/Screening (PGD/PGS)

MONASH IVF GROUP

Key points:

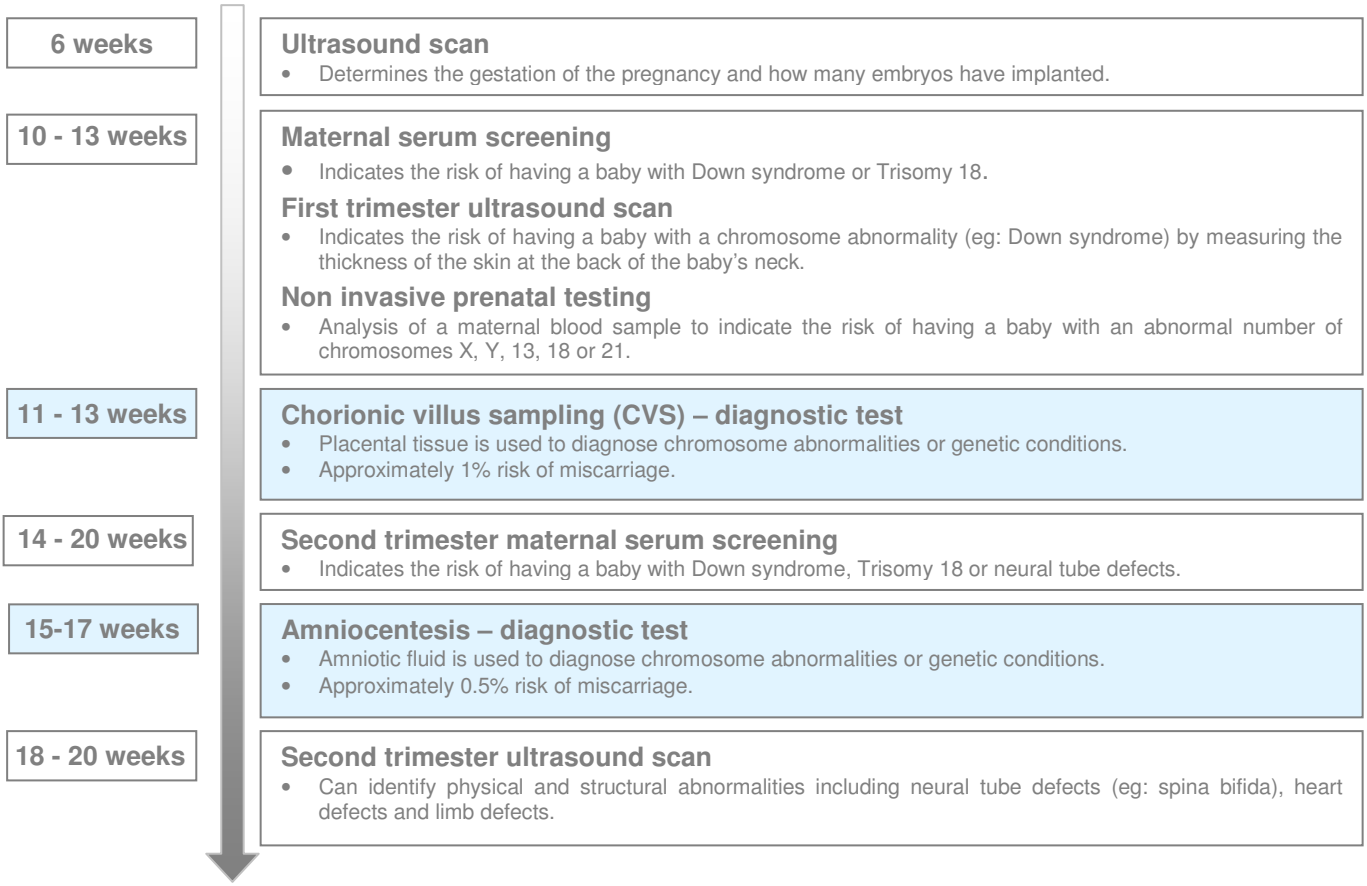
- PGD/PGS is a screening test and is not 100% accurate. Confirmatory prenatal diagnosis is highly recommended if a pregnancy is achieved following PGD/PGS.
- There are a number of different prenatal tests available to assess the development of the baby during the pregnancy. Some of these prenatal tests are screening tests (eg: ultrasound, maternal serum screening), while others are diagnostic tests (eg: Chorionic Villus Sampling and Amniocentesis).
- A prenatal diagnostic test (eg: Chorionic Villus Sampling or Amniocentesis) is highly recommended for couples who have had PGD/PGS in order to confirm the PGD/PGS result.

What is confirmatory prenatal diagnosis following PGD?

Preimplantation Genetic Diagnosis (PGD) and Preimplantation Genetic Screening (PGS) are techniques that enable IVF embryos to undergo genetic screening before a pregnancy is established. The results obtained from PGD and PGS are not 100% accurate. The accuracy of the result can range from 90% to 99% depending on the type of test being performed, the quality of the biopsy sample and the clarity of results obtained. Consequently, there may be up to a 10% error rate associated with any test. It is recommended that all patients who achieve a pregnancy following PGD/PGS undergo a diagnostic prenatal test in order to confirm the PGD/PGS result.

There are lots of different prenatal testing options available during pregnancy (Figure 1). These tests are typically classified as screening tests (which indicate whether a pregnancy is at risk of a genetic condition) or diagnostic tests (which provide a definitive diagnostic result). Confirmation of a PGD/PGS result requires a diagnostic test.

Figure 1: Prenatal testing options available during pregnancy. The shaded boxes represent diagnostic tests while the white boxes represent screening tests.



How are diagnostic tests done?

There are two different diagnostic tests that can be performed during pregnancy to confirm the PGD/PGS result, namely Chorionic Villus Sampling (CVS) or Amniocentesis. Each of these procedures is discussed in more detail below.

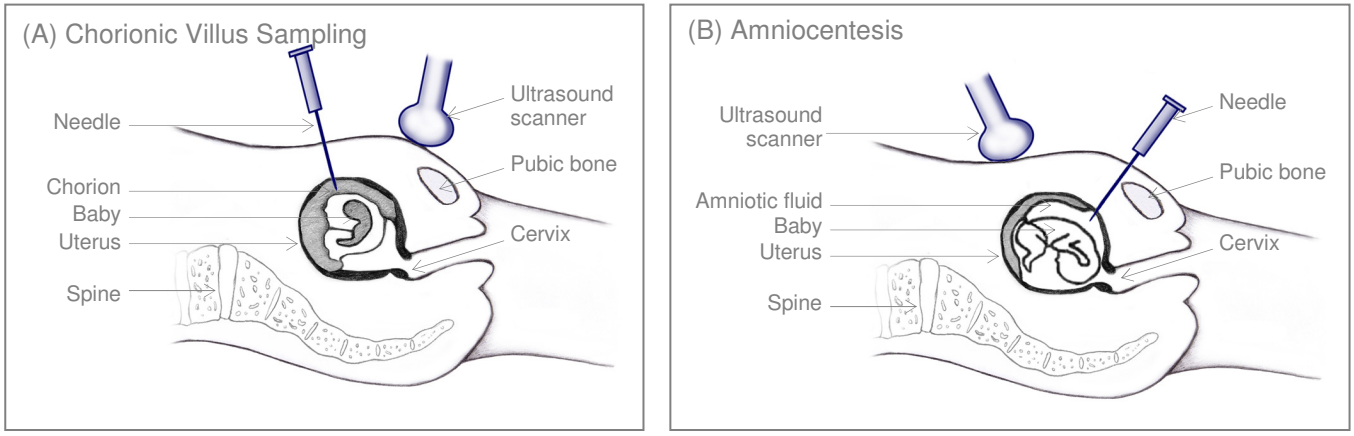
Chorionic villus sampling

CVS is usually performed between 11 and 13 weeks of gestation (Figure 2A). A local anaesthetic is used to numb the area of skin where the sampling needle will be inserted. A thin needle is inserted through the abdomen and into the womb. The needle is observed at all times during the procedure using ultrasound to ensure that it does not go near the baby. A small sample of the placental tissue (chorionic villi) is removed for genetic analysis. Cells from the placental tissue usually have the same genetic makeup as the baby. There is an approximate 1% risk of miscarriage following CVS. In rare cases, the CVS result may be difficult to interpret or a result may not be provided. In these cases an amniocentesis is offered to clarify the results.

Amniocentesis

Amniocentesis is usually performed between 15 and 17 weeks of gestation (Figure 2B). A local anaesthetic is used to numb the area of skin where the sampling needle will be inserted. A thin needle is inserted through the abdomen and into the womb. The needle is observed at all times during the procedure using ultrasound to ensure that it does not go near the baby. A small sample of amniotic fluid (which contains cells shed by the fetus) is removed for genetic analysis. There is an approximate 0.5% risk of miscarriage following amniocentesis. In rare cases, a result may not be obtained and a repeat sample may need to be taken.

Figure 2: Prenatal diagnosis options.



What to do next?

It is recommended that you contact your local Genetic Counselling Service (using the details provided on the next page) to discuss prenatal diagnostic testing in more detail. This is an important step to ensure you are fully informed about your prenatal testing options and the associated benefits, risks and limitations.

Genetic counselling service contact details

Victoria		
Metropolitan Services		
Clayton (Monash Medical Centre)	(03) 9594 2026	
Parkville (The Women's Hospital)	(03) 8345 2180	
Regional Services	(03) 8341 6297 Ballarat, Bendigo Geelong, Mildura, Warrnambool (03) 8341 6332 Gippsland	
Australian Capital Territory	(02) 6244 2133	
New South Wales	(02) 9845 3273 The Children's Hospital at Westmead (02) 6041 3545 Albury/Wodonga	
Queensland	(07) 3636 1686	
South Australia	(08) 8161 7375	
Tasmania	(03) 6222 8296	
Western Australia	(08) 9340 1525	
New Zealand		
Auckland	(09) 307 4949 Extn: 25870	Toll Free: 0800 476 123
Wellington	(04) 385 5310	Toll Free: 0508 364 436
Christchurch	Ph: (03) 378 6574	Toll Free: 0508 364 436