

What do you know about Prenatal Diagnosis?



衛生署
Department of Health



HOSPITAL
AUTHORITY
醫院管理局

What do you know about Prenatal Diagnosis?

Prenatal diagnosis aims at detection and appropriate management of severe diseases of the fetus. Management options may include:

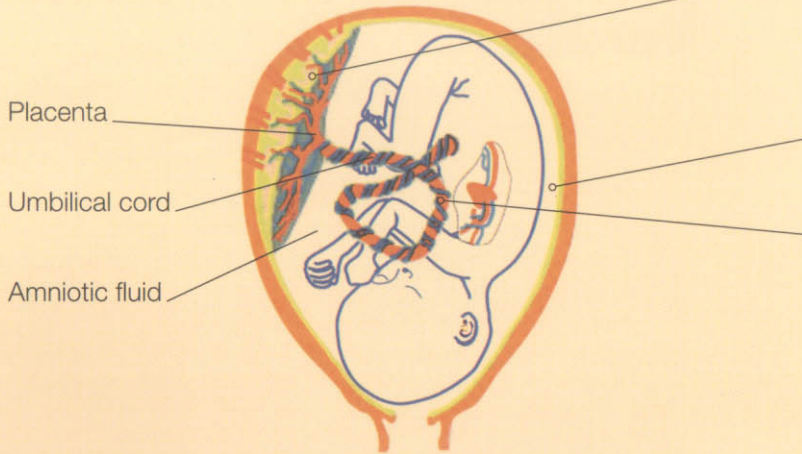
- **Direct treatment of the fetus inside womb**
- **Preparations for optimal management after birth**
- **Termination of pregnancy**

Prenatal diagnosis involves testing pregnant women at high risk of having babies with major congenital abnormalities or hereditary conditions. These include:

- Older women who have higher chance of having chromosomal abnormal babies like Down's syndrome.
- Women who have given births to abnormal babies with congenital heart diseases, spinal defects, or other birth lesions.
- Women with family history of genetic diseases, such as haemophilia.
- Women with partners both have thalassernia trait.
- Women who may be considered to be at risk based on clinical assessment by their obstetricians.

Certainly, there is limitation that not all abnormalities and diseases could be detected by our current medical technology.


What do you know about Prenatal Diagnosis?





The method used is related to the type of abnormalities being considered. There are two main types of tests: invasive tests to obtain fetal cells or fetal related cells for chromosomal and other analyses and imaging to detect structural abnormalities.

a) Invasive tests

For chromosomal abnormalities, direct culture of cell samples from the fetus by one of the following methods is performed at different gestational age: chorionic villus sampling, amniocentesis or cordocentesis.

 **Chorionic villus sampling** - It is performed between 10 to 13 weeks of gestation. A sample of placental tissue is obtained and the results of chromosomal analysis are usually available in 2-3 weeks.

 **Amniocentesis** - It is usually performed between 16 to 20 weeks of gestation. A sample of liquor surrounding the fetus is taken. Results of chromosomal analysis are usually available within 3 weeks.

 **Cordocentesis** - It is usually done after 20 weeks of gestation. A sample of fetus blood is taken from the umbilical cord. The results of chromosomal analysis are usually available in 5- 7 days. *The test is particularly useful if an urgent result is desired.*

How are these tests performed?

All these procedures are performed under ultrasound guidance. A long needle is used to obtain the tissue sample, which is subsequently cultured in the laboratory. Chromosomal abnormalities and some genetic or hereditary conditions such as thalassemia major can then be diagnosed or excluded.

Is the result reliable?

Chromosomal analysis using the above methods is highly accurate. However, expert interpretation by specialist is mandatory because some chromosomal abnormalities are just normal variations and will not result in deficient physical or mental growth of the fetus.

Will these tests cause harms to the fetus?





These tests are associated with a definite but small risk of miscarriage of around 0.5 to 1.5% under the care of expert doctors, i.e. 1-3 out of 200 women receiving the invasive test will result in miscarriage.

If the result is normal, does it mean that the fetus is normal?

Not all genetic or hereditary diseases can be detected by these methods due to limitations in medical technology.

Structural defects or functional defects not related to chromosomal abnormalities will not be detected.

b) Ultrasound examination

-  The most useful test for detection of major structural abnormalities in the fetus is ultrasound examination.
-  **The detection rate for major structural abnormalities by ultrasound examination between 18 to 22 weeks gestation is around 30 - 70 %**, the rate being different for different abnormalities or organ systems concerned.
-  Experience of the doctor performing ultrasound as well as the resolution of the ultrasound machine affect the detection rate.
-  If the abnormalities detected on ultrasound examination are suggestive of chromosomal disorders, further invasive testing as mentioned above may be indicated.



If ultrasound examination is normal, does it mean that the fetus is normal?

- While a normal ultrasound examination implies that the chances of major structural defects in the fetus should be small, it does not exclude all possibilities of such defects.
- Some chromosomal abnormalities, may not be readily detectable by ultrasound, so a normal ultrasound examination does not exclude these diseases.

How and where to get these prenatal diagnostic tests?

All pregnant women are encouraged to discuss with their doctors if they have genuine concerns that their fetuses could be abnormal.

Prenatal diagnosis and counselling service is available at all major obstetric units under the Hospital Authority. Couples requesting prenatal diagnostic and counselling services should approach the Hospital Authority hospitals with obstetric services in their districts, or they can be referred to one of these clinics through their own doctors, from Maternal and Child Health Centres, or other private clinics.