What Do You Know about Prenatal Diagnosis?

Prenatal screening test can assess your risk (or chance) of having a baby with severe congenital condition and enable early management. In addition, the Hospital Authority offers prenatal screening for Down Syndrome according to the gestation of pregnancy.

What is prenatal diagnosis?

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

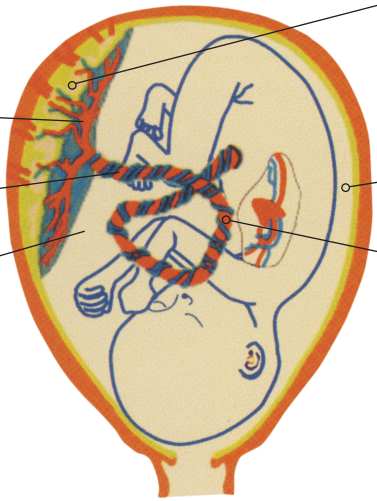
- Direct treatment of the foetus inside the womb
- Preparations for optimal management after birth
- Termination of pregnancy

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

- 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.
- Couples who are both thalassaemia carriers.
- Women who are considered to be at risk based on clinical assessment by their obstetricians.

Prenatal diagnostic tests have limitations and not all abnormalities and diseases can be detected by the 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: (A) invasive tests to obtain foetal cells or foetal related cells for chromosomal and other analyses and (B) imaging to detect structural abnormalities.

(A) Invasive tests

For chromosomal abnormalities, direct culture of cell samples from the foetus 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 result of chromosomal analysis is usually available in 2-3 weeks.

Amniocentesis –
It is usually performed between 16 to 20 weeks of gestation. A sample of liquor surrounding the foetus is taken. Result of chromosomal analysis is usually available within 3 weeks.

Cordocentesis –
It is usually done after 20 weeks of gestation. A sample of foetal blood is taken from the umbilical cord. The result of chromosomal analysis is 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 inside uterus, which is subsequently cultured in the laboratory. Chromosomal abnormalities and some genetic or hereditary conditions such as thalassaemia 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 foetus.

Will these tests cause harms to the foetus?

Some invasive 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 foetus 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 foetus is ultrasound examination.
- **The detection rate for major structural abnormalities by ultrasound examination done between 18 to 22 weeks gestation is around 30-70%.**
- 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 test as mentioned above may be indicated.

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

- **While a normal ultrasound examination implies that the chances of major structural defects in the foetus 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 foetus 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, Maternal and Child Health Centres, or other private clinics.

(This leaflet is prepared by the Department of Health and the Hospital Authority)

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