What are the possible test results?

There are three possible test results:

(The interpretation of results is based on information available at the time of reporting.)

1. **Normal**
   No clinically significant gain or loss of genetic material is detected in the fetal chromosome.
2. **Pathogenic**
   Gain or loss of genetic material is detected in the fetal chromosome. The clinical features associated with these results depend on the specific genetic material that is gained or lost.
3. **Uncertain clinical significance**
   Gain or loss of genetic material is detected in the fetal chromosome but the effect on the fetus is uncertain. Performing CMA test on the blood samples taken from both biological parents may help in interpretation.

What are the limitations of this test?

* cannot detect balanced structural arrangement of chromosomes.
* cannot detect individual gene changes and regions with no probe coverage
* cannot identify uniparental heterodisomy or small region of absence of heterozygosity.
* cannot detect low level mosaicism (presence of cells with different chromosomal makeup in the baby or the placenta).
* a normal test result cannot exclude all abnormalities.

What are the other important considerations in choosing this test?

* It is possible that a diagnosis unrelated to the reason of testing may be found including predisposition to mental retardation, autism, cancer, late-onset diseases or other medical conditions.
* There is a small chance of finding a genetic condition affecting the health of yourself or other family members.
* The test result may still be uncertain even after parental blood is tested.
* The above may impose psychological distress.
* You need to indicate what information you would not like to know from the test.

Charge

Please ask your doctor for the price of the following testing:

CMA

CMA+ QF-PCR

CMA+ QF-PCR + karyotyping

The above service is provided by Mrs Wu Chung Prenatal Diagnostic Laboratory of Tsan Yuk Hospital.

Please ask your doctor if you have any questions about the information in this brochure.

Please visit http://www.obsgyn.hku.hk/prenatal_diagnosis for more information

V5-1906

Prenatal Chromosomal Microarray (CMA) Testing

Prenatal Diagnostic and Counselling Division

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Department of Obstetrics & Gynaecology
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What are the objectives and benefits of prenatal CMA testing?

CMA (chromosomal microarray) is a special test which finds differences in the amount of genetic material in an individual. It looks for areas of the chromosomes that have gain or loss of genetic material.

The usual examination of chromosomes under the microscope is called karyotyping.

CMA test is much more sensitive than karyotyping. CMA has the potential to find a chromosome gain or loss even if testing of the chromosomes by karyotyping shows normal results.

These smaller imbalances, possible gains or losses of genetic material, are called 'submicroscopic imbalances' because they cannot be seen through the microscope. They may cause birth defects, delays in development, and genetic syndromes.

SNP array, a form of CMA, can detect regions of absence of heterozygosity (AOH), which is usually caused by an abnormal inheritance pattern (Uniparental Disomy, UPD) of having a pair of chromosome from just one parent (mother of father). In some instances, UPD may result in a genetic disorder and increase chance of having recessive genetic disease due to AOH.

It is a fast test. The result can be available in 7 working days. In contrast, conventional chromosome testing by karyotyping takes 3 weeks to provide a result.

An early prenatal diagnosis of a gain or loss of genetic material on a specific chromosome can provide additional information that may enable your doctors to manage your pregnancy better and also may enable you and your doctors to know what to expect after delivery of the baby.

Who should consider having this test?

Women who are at increased risk of having babies with congenital abnormalities may choose to have the test when prenatal invasive diagnostic procedures are performed. Common reasons for invasive diagnostic procedures include abnormal ultrasound findings of the baby, previous history of an abnormal baby, family history of a genetic disorder, or if Down syndrome screening test indicates an increased risk for a chromosome abnormality. Women with stillbirth may also benefit from the test to evaluate the cause of fetal death.

What types of samples are required for this test?

* Amniotic fluid or chorionic villi obtained at amniocentesis or chorionic villus sampling (CVS). Placental tissue or skin biopsy from stillbirth.
* 3 mL blood from both biological parents^, wherever possible, at the time of sampling.

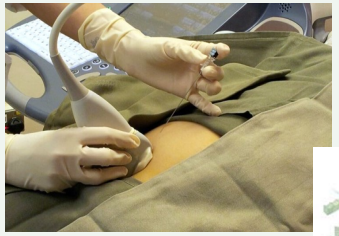
^ CMA is a very sensitive test which may identify genetic changes which are inherited from either parent of the baby and may not affect the baby's health. Therefore, it is important that both biological parents provide a blood sample for analysis and interpretation. Parental samples will not be further processed if the CMA result for the fetal sample is normal, hence no parental report.

How is the test done?

The DNA of the fetus is taken from amniotic fluid cells or chorionic villi. Microarray analysis is performed using a SNP based Affymetrix CytoScan 750K array.

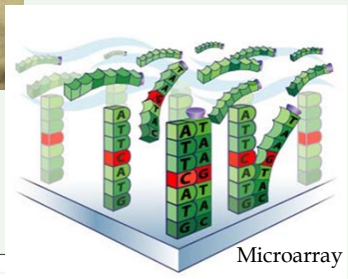
When will the results be available?

* If the sample is adequate and the result normal, the fetal test result will be reported in 7 working days.
* If a chromosomal problem is suspected, tests to confirm these findings will be performed. Depending on the complexity of the problem, the report will be available in 2-3 weeks or more.

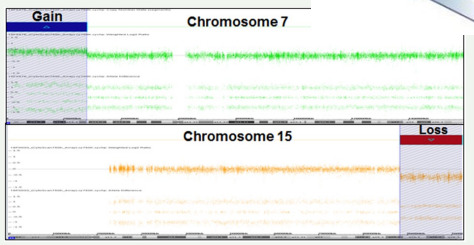


Amniocentesis under ultrasound guidance

Sample DNA



Microarray



How do I get to know the results?

* The test result will be reported to your doctor, who will explain the result to you.
* You and your family members may be referred to clinical geneticist for further counselling if there are abnormal findings (pathogenic or uncertain clinical significance).