 瑪麗醫院 QUEEN MARY HOSPITAL	<b>Department of Obstetrics and Gynaecology</b>	<b>Document No.</b>	<b>OGPD0011(I)-E</b>
	<b>Subject</b> Information on Chromosomal Microarray Testing (Postnatal)	<b>Issue Date</b>	<b>Jan 2025</b>
		<b>Review Date</b>	<b>Jan 2028</b>
		<b>Approved by</b>	<b>Prenatal Diagnosis and Counselling Team, TYH</b>
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## **Information on Chromosomal Microarray (CMA) Testing (Postnatal)**

### **What are the objectives and benefits of CMA testing?**

- Chromosomal microarray (CMA) is a special test which finds differences in the amount of genetic material among individuals. 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 or father). In some instances, UPD may result in a genetic disorder and increase chance of having recessive genetic disease due to AOH.

### **Who should consider having this test?**

- CMA is the recommended test for individuals with developmental or congenital problems. The clinical geneticist will assess if the test is necessary for your child.

### **What type of samples is required for this test?**

- 3 mL blood from the patient.

### **How is the test done?**


- The DNA of the patient is taken from blood. Microarray analysis is performed using a SNP based Affymetrix CytoScan 750K array.

### **When will the results be available?**

- The test will be reported in 21 days.

### **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).

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**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 chromosome.
2. **Pathogenic** – gain or loss of genetic material is detected in the 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 chromosome but the effect 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 an individual).
- 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 problems, 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.

*I acknowledge that the above information has been discussed with me by the medical staff and I fully understand the information. I have been given the opportunities to ask questions pertinent to the test and satisfactory answers have been provided by medical staff.*

GUM LABEL

Signature \_\_\_\_\_

Date \_\_\_\_\_