CONSENT FORM

PRENATAL CHROMOSOME MICROARRAY ANALYSIS	ACCOUNT NO. NRIC NO. NAME ADDRESS BIRTH DATE/RACE DATE AND TIME OF ADMISSION

What is Chromosome Microarray Analysis (CMA)?

Human bodies are made up of millions of cells. Each cell contains our genetic material, or DNA. The complete set of all our genetic material is called our genome. **Chromosome microarray analysis (CMA)** is a genetic test that looks for extra or missing DNA materials in a person's genome.

There are usually two copies of each gene in our cells. If a person has more than or less than two copies of a particular gene, this may result in problems such as functioning and development of that individual.

Why do you need this test and what are its limitations?

You may be offered prenatal CMA if there are abnormal ultrasound findings in your pregnancy, abnormal first trimester screening results, or if there are some other high-risk indications for the test.

CMA has greater sensitivity than the conventional chromosomal analysis. It can detect abnormalities that would not have been previously identified from chromosomal analysis. If an abnormality is identified, the results may provide important information that can be used to guide medical decisions and/ or future reproductive decisions.

Although CMA is a very sensitive test, it only detects changes in the number of DNA copies. It cannot detect many other conditions in which there is a change, or a mutation, in a gene, but the number of DNA copies remains the same.

What does it involve?

A sample obtained via amniocentesis (sampling of amniotic fluid), chorionic villus sampling or cordocentesis (sampling of umbilical cord blood) is needed for prenatal CMA.

Additional material, in the form of blood samples from parents or other family members, may be requested to determine if additional DNA copies (also known as copy number variations, CNVs) detected is/are inherited from the parents or is/are present in the fetus only. This will help towards interpretation of results.

The indication(s) for performing CMA, the turnaround time and its associated costs will be explained to you, and the indication(s) will be provided to the testing laboratory. This clinical information is required for accurate interpretation of results.

What precautions must you take for this test?

Please inform your doctor of the following as it could affect the accuracy of the results:

- If you have received bone marrow transplant or recent blood transfusion.

- If the pregnancy is as a result of egg, sperm or embryo donation.
- If you or your partner are closely related biologically.

What are the concerns of the test?

- The interpretation of the test results is based on the necessary clinical information which the laboratory has access to, as at the date of issuance of the report.
- A "normal" prenatal CMA report (no clinically significant CNVs detected) does not exclude all genetic conditions in the fetus being tested. It does not guarantee that the fetus being tested will be healthy, or will not develop diseases later in life.
- All genetic tests have limitations. False negative and false positive results, though rare, may occur.
- Additional material may be required if the sample collected is insufficient, damaged in handling or not viable.
- CMA detects virtually all the established cytogenetically defined microdeletions and microduplications greater than 400 kb. However, some CMA gains or losses may be associated with phenotypes that vary from normal to severe. It may not be possible to predict what the phenotype will be in a particular individual, and may not be reported.
- As Prenatal CMA looks at targeted regions of the genome covering almost the entire genome, it can generate a lot of data from one test. Although most of this information is useful, the data can be difficult to interpret, and can lead to results that are of uncertain or of unknown significance. Further confirmation test may be required to ascertain if the variation in genome is clinically significant
- Sometimes, CMA testing may also reveal unexpected results, particularly for conditions that appear later in life, which a person may not want to know about. Some families find it difficult to deal with these results.
- Genetic testing of parents may reveal information about yourself, your fetus or your family. For example, the test may reveal non-paternity, the use of an egg or sperm donor, or that the parents are closely related biologically.
- Genetic test results may result in some forms of discrimination (insurance, employment or other) as they form part of your medical record and may be accessed by and/or disclosed to a third party who has obtained my necessary consent or when such access is otherwise allowed or required by law.
- The test results may reveal incidental findings not related to the original indication for the test. Please discuss this further with your clinician.

What will happen to your test results and sample once the test is complete?

Due to the complexity of the test, your results will only be made available to you by your ordering clinician, genetic counsellor or suitably qualified and appointed healthcare professional.

After testing is completed, the unused sample may be destroyed or anonymised, stored and used for laboratory internal validation, quality control and process development. When anonymised, any results obtained cannot be traced to the original source and no further results will be reported. In all instances, the sample will be handled according to laboratory retention and storage policy. No other tests other than those authorised will be performed. Your

information and records will be maintained according to the hospital's standard policies on medical confidentiality.

What are your options?

Participation in the test is voluntary. You can withdraw from the test before completion of the test, you can do so by requesting your ordering clinician to inform the laboratory but you will be charged for the full cost of the test.

Others (to be filled by Medical Practitioner) [if applicable]

Part I – Patient's Declaration

1. I, _____ (NRIC/Passport No. _____), have read this information sheet and confirm that I understand the nature, purpose, concerns, and limitations with regard to **Prenatal Chromosome Microarray Analysis** ("Test").
2. I understand and agree that the Test will be performed by the appropriate SingHealth institution (with the involvement of external service providers if necessary) and I will be admitted and/or registered as a patient of that SingHealth institution ("Hospital").
3. I agree and give permission for my clinical details, sample(s) or derivatives, and any personal data set out in the accompanying test order form to be used by my ordering clinician and/or the Hospital for performing the Test and any necessary follow-up actions.
4. I acknowledge that the concerns listed are not intended to be exhaustive. I have had an opportunity to ask for more information about (i) the above-mentioned concerns; (ii) the concerns in general; and (iii) specific concern(s) of relevance to me during pre-test genetic counselling.
5. I hereby consent to the Test.

_____ _____

(Signature/**[*Left/Right]** Thumbprint of Patient) (Date of Signing)

_____ _____

(Name of Witness) (Designation of Witness)

_____ _____

(Signature of Witness) (Date of Signing)

*** Please delete accordingly**

Part II – Parent's / Legal Guardian's / Donee's / Deputy's Declaration (herein referred to as the "Authorised Person") (if applicable)

1. I, _____ (NRIC/Passport No. _____), the ***Parent / Legal Guardian / Donee / Deputy** of _____ (NRIC/Passport No. _____) ("Patient"), have read this information sheet and confirm that I understand the nature, purpose, concerns, and limitation with regard to **Prenatal Chromosome Microarray Analysis** ("Test").
2. I understand and agree that the Test will be performed by the appropriate SingHealth institution (with the involvement of external service providers if necessary) and the Patient will be admitted and/or registered as a patient of that SingHealth institution ("Hospital").
3. I agree and give permission for the Patient's clinical details, sample(s) or derivatives, and any personal data set out in the accompanying test order form to be used by the Patient's ordering clinician and/or the Hospital for performing the Test and any necessary follow-up actions.
4. I acknowledge that the concerns listed are not intended to be exhaustive. I have had an opportunity to ask for more information about (i) the above-mentioned concerns; (ii) the concerns in general; and (iii) specific concern(s) of relevance to me during the pre-test genetic counselling.
5. I hereby consent for the Patient to undergo the Test.

_____	_____
(Signature/**[*Left/Right]** Thumbprint of Authorised Person)	(Date of Signing)
_____	_____
(Name of Witness)	(Designation of Witness)
_____	_____
(Signature of Witness)	(Date of Signing)

*** Please delete accordingly**

Part III – Medical Practitioner's Declaration

I confirm that I have explained to the Patient, or the Authorised Person (if applicable), the Patient's medical condition as well as the nature, purpose, concerns, and limitations with regard to the Test and have addressed queries of the Patient, or the Authorised Person (if applicable).

_____	_____
(Full Name, MCR Number, and Signature of Medical Practitioner)	(Date of Signing)

Part IV – Interpreter's Declaration (if applicable)

I, _____confirm that I have interpreted to the Patient, or the Authorised Person (if applicable), the Medical Practitioner's explanation of the Patient's medical condition, nature, purpose, concerns, and limitations with regard to the Test and the Medical Practitioner's response to the Patient's, or the Authorised Person's (if applicable), queries in _____ (language / dialect).

_____	_____
(Signature of Interpreter)	(Date of Signing)