# CONSENT FORM

| **KARYOTYPING / FLUORESCENCE IN SITU HYBRIDISATION (FISH) FOR CONSTITUTIONAL GENETIC TESTING** | ACCOUNT NO.<br>NRIC NO.<br>NAME<br>ADDRESS<br>SEX/BIRTH DATE/RACE<br>DATE AND TIME OF ADMISSION |
|---|---|

## What is karyotyping / Fluorescence in situ Hybridisation (FISH)?

**Karyotyping** is the study of the chromosomes that are present in human cells under the light microscope. The chromosomes are structures on which the genes are located.

**FISH** is a specialized technology that uses fluorescent labelled DNA fragments that bind to a specific sequence on the chromosomes.

## Why do you need this test(s) and what are its limitations?

**Karyotyping** can detect abnormalities in chromosome number (such as trisomy 21), large deletions/duplications of chromosomal material and balanced chromosomal rearrangements. This information can lead to a specific diagnosis. Occasionally, a structural abnormality may not be detectable as it is too small to be seen visually.

**FISH** can provide clinical information on chromosomal abnormalities that are too small to be detected by standard karyotyping. This information includes subtle deletions/duplications and rearrangements. This test is highly accurate in appropriate applications, but since some disorders may have more than one cause or is associated with deletions or duplications beyond the capability of the FISH analysis, FISH may not suffice for diagnostic purposes.

## What does it involve?

The test can be performed on a sample of blood, tissue, bone marrow, chorionic villi or amniotic fluid. All procedures in sample collection carry their associated risks and these risks have been explained to you.

Additional material, in the form of blood samples from you, your parents or other family members, may be requested to determine whether or not a detected chromosomal aberration/variant is inherited. This will help to determine if the chromosomal aberration/variant found in you is clinically significant.

The indication(s) for performing karyotyping/FISH, the turnaround time of the test 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 the test(s)? (if applicable)

Please inform your clinician if you have received a bone marrow transplant or recent blood transfusion.

## What are the concerns of the test(s)?

- 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” karyotype report does not exclude all genetic conditions in the subject being tested. It does not guarantee that the subject being tested will be healthy, or will not develop genetic 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.
- Genetic test results may result in some forms of discrimination (insurance, employment or others) 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(s) results and sample once the test(s) is/are completed?**

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, your 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(s) is voluntary. If you wish to 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 regards to the following test(s) ("Test(s)"):

   ☐ **Karyotyping**

   ☐ **Fluorescence in situ Hybridisation (FISH)**

2. I understand and agree that the Test(s) 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(s) and any necessary follow-up actions.
4. I acknowledge that the concern(s) 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(s).

_____ _____

(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 _____ (Birth Certificate/NRIC/Passport No. _____) ("Patient"), have read this information sheet and confirm that I understand the nature, purpose, concerns, and limitations with regards to the following test(s) ("Test(s)"):

   ☐ **Karyotyping**

   ☐ **Fluorescence in situ Hybridisation (FISH)**

2. I understand and agree that the Test(s) 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(s) 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 the Patient during the pre-test genetic counselling.
5. I hereby consent for the Patient to undergo the Test(s).

_____ _____

(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(s) 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(s) and the Medical Practitioner's response to the Patient's, or the Authorised Person's (if applicable), queries in _____ (language / dialect).

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