

Affix Patient Label

- ☐ National University Hospital  
☐ Ng Teng Fong General Hospital  
☐ Alexandra Hospital  
☐ Jurong Community Hospital  
☐ Jurong Medical Centre

From

## Genetic Testing Consent Form

### PART I – NAME OF TREATMENT/ OPERATION/ PROCEDURE

#### Procedure Specific Description

Test ordered by your provider:

- a) Specific Variant testing ☐ b) Gene Panel ☐ c) Whole Exome Sequencing / Whole Genome Sequencing ☐  
d) Chromosomal analysis ☐ e) Carrier screening ☐ f) Others \_\_\_\_\_

Name of Test: \_\_\_\_\_

#### 1. Purpose of Genetic Testing

The purpose of genetic testing is to analyse your genetic material to identify changes (also called variants) that may contribute to your health or your family's health.

#### 2. The potential benefits of genetic testing

The potential benefits may include:

- Confirming or clarifying a genetic diagnosis
- Enabling your doctors to give you more personalised medical advice and care
- Determining whether you are a carrier of a genetic condition, which may affect your risk of passing on a genetic condition to your children
- Estimating your risk of developing specific genetic health conditions in the future
- Helping your family members understand and identify if they may be at risk for certain genetic health conditions

#### 3. Important Pre-Test Considerations

- It is important to inform your doctor or genetic counsellor if you have received a recent blood transfusion or had previously undergone bone marrow transplant as these may affect what test is done, the sample used, the results of your genetic test etc

#### 4. The Testing Process

##### **Sample Collection:**

A sample will be collected from you. This may include a blood sample, saliva, buccal (cheek) swab, hair follicle, or other tissue (please specify: \_\_\_\_\_). The type of sample required depends on the specific test being performed.

##### **Laboratory Analysis:**

Your sample will be sent to a certified genetic laboratory, where it will be processed and analysed.

##### **Scope of Testing:**

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Only the genes or genetic variants that you consented to will be analysed. No additional genetic testing will be done without your separate, explicit consent. If you request further testing or reanalysis in the future, this may require an additional sample and may result in extra charges.

**Accuracy and Interpretation:**

Genetic testing is generally highly accurate in detecting genetic changes. However, some variants may not be identified due to technical limitations or the current state of scientific and medical knowledge. The genetic changes are interpreted using the most up-to-date information, but some changes may be uncertain or reclassified as new research becomes available.

**After Testing:**

Due to the complexity of the genetic test and its results, you will meet up with a genetic specialist, genetic counsellor or suitably qualified healthcare professional. They will inform you about what your results and what it means for your health and, if applicable, for your family members. They may recommend relevant follow up care or tests.

**Sample Handling and Storage:**

Any remaining portion of your sample will not be returned to you. It may be stored by the laboratory for a limited time and used for process improvement or quality assurance, in accordance with patient confidentiality policies and regulatory guidelines.

**5. Limitations and Risks of Genetic Testing**

The potential limitations and risks are

- The genetic test result may confirm a genetic diagnosis, but may not be able to predict the severity, progression, or exact timing of health complications.
- Specific therapies for the identified genetic condition may or may not be available.
- The genetic test result may be associated with increased risk for certain forms of discrimination, such as for insurance or employment.
- The genetic test result may have implications for the health risks of your biological family members.
- There is a small chance of error in the results e.g. due to technical limitations, sample-related issues, differences in variant classification, inaccurate or incomplete family or clinical history, or other scientific limitations.
- Occasionally, the laboratory may require an additional sample to clarify your test result.
- In rare cases, genetic testing may reveal unexpected information about family relationships, such as non-paternity or undisclosed adoption
- People may react differently to receiving genetic test results. Some may feel stressed, anxious, sad etc. If needed, you can request additional support or counselling before or after receiving your results.

**6. Test specific information:**

**a. Specific Variant testing**

- This examines only a single, predetermined genetic variant that has been identified as relevant to your clinical situation (for example, a variant previously found in a family member).
- No analysis of other genes or variants will be performed without your consent

**b. Gene panel testing**

- This analyses a defined group of genes that are known to be associated with a particular medical condition or set of symptoms.
- Only the selected genes of that test will be analysed. No broader genetic analysis will be conducted without your consent.

**c. Whole Exome Sequencing/Whole Genome Sequencing (WES/WGS)**

- Whole Exome Sequencing (WES) looks at the parts of your DNA that provide instructions for making proteins (called exons). These regions are where most known disease-causing genetic changes are found.

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- Whole Genome Sequencing (WGS) looks at your entire DNA, including both the protein-coding regions and the parts that do not code for proteins.
- Testing can be patient only or patient and family members. If your test includes samples from your parents or other close relatives ("trio" or "duo" testing), their samples will only be analysed to help interpret your results. Any findings directly related to your health and the reason for testing will be reported. Family members will not receive individual results from this test.
- This test does not include a full carrier screening for recessive genetic conditions that are not related to the reason for your test.
- Secondary Findings/Incidental Findings
  - Genetic testing may discover other genetic changes unrelated to the reason you are being tested.
  - In some cases, finding out about these changes can allow you to take steps for early diagnosis, intervention, or better management of possible health conditions.
  - In other cases, we may find out information such as non-paternity, undisclosed adoption
  - You can choose whether you want to receive information about these additional findings, if any are discovered.
- d. **Chromosomal analysis (Karyotyping/Fluorescence in situ hybridization/Chromosomal microarray/Low pass genome sequencing for aneuploidy)**
  - Chromosomal analysis is done to see if there is any variation in your chromosomes.
  - There are different types of tests that can analyse chromosomes.
  - Chromosomal karyotyping/fluorescence in situ hybridization can detect abnormal chromosome numbers, large rearrangements, large deletions and duplications. However, it has a limited resolution and cannot detect other genetic changes such as small deletions, small duplications, and point mutations. Chromosomal karyotyping requires mitotic (dividing) cells from a live specimen; the test may not be successful if the cells are not healthy.
  - Chromosomal microarray or low pass genome sequencing for aneuploidy can detect abnormal chromosomal numbers, large and small deletions, as well as large and small duplications. It cannot detect point mutations. It cannot detect large rearrangements when this is no change in DNA copies.
  - In the process of chromosomal analysis, there may be unexpected test results not directly related to the primary reason for performing the test.
- e. **Carrier Screening**
  - Carrier screening is a test that looks at specific genes to find out if you carry a genetic change for certain inherited conditions. Even if you do not have any symptoms, you can still be a carrier and pass these conditions to your children.
  - This test helps couples understand their chances of having a child with a genetic disorder. Knowing your results can help you make informed choices about family planning.
  - Carrier screening will have positive or negative test results only. Variants of uncertain significance will not be reported.
  - Positive result:
    - This means a genetic change was found
    - Most of the time, this means you are a carrier of a genetic disorder
    - In some cases, if more than one genetic change is found in the same gene, you might be affected by the disorder or have a risk of developing it in the future.
  - Negative result:
    - This means no known disease-causing genetic changes was found
    - A negative result indicates that no genetic change was identified. This reduces but does not eliminate the risk of being a genetic carrier as there may be genetic changes undetected by this test due to limitations in current technology or knowledge.
- f. **Other genetic test**
  - (Free text to specify e.g. MsPCR for Russell Silver/Beckwith Wiederman syndrome etc)

## 7. **Possible outcomes of genetic testing**

The possible outcomes of genetic testing include:

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- **Positive, Genetic variant(s) identified**
  - This means a genetic change related to your condition was found
  - This result confirms that you either have the genetic condition, are a carrier, or are at increased risk for the disease.
  - This information can help guide your medical care and may also be important for your family members to know about their own health or risk.
- **Negative, Genetic variant(s) not identified**
  - No genetic changes linked to your condition were found in this test.
  - This may or may not completely rule out a genetic cause for your condition. This depends on the indication for the test, the condition that you are being tested for and the current knowledge.
  - Your healthcare team will explain this further to you.
- **Uncertain, Variant(s) of uncertain significance (VUS) identified**
  - A genetic change was found, but we do not yet know if this change causes disease or affects your health
  - This result will not be used to direct medical management, unless deemed so by your doctor.
  - Sometimes, testing other family members can help us learn more about the significance of this result.
  - As scientific understanding improves, these uncertain results may be reclassified. If this happens, the laboratory may update the report and let you or your healthcare provider know.

#### **Data storage and confidentiality**

The laboratory and healthcare institution will take all reasonable steps to protect your privacy in compliance with applicable data protection laws and regulations.

Your genetic test sequence and result(s) will be stored as part of your records, which will be accessible by the medical teams involved in your care, including at your GP or polyclinic, and will only be released for the purposes set out in this form or as allowed under law.

Your information may be collected, used, and disclosed by public healthcare entities and public agencies in relation to the provision of financial subsidies and healthcare services, and for policy and services planning and analysis.

Your de-identified genetic and clinical information may be added to and used in healthcare or scientific databases to facilitate more accurate diagnoses, support medical research, develop new treatments, and improve the capability to predict and prevent diseases.

Your result(s) may be useful to your family members to receive genetic counselling and testing.

Please see our PDPA notice online at [<https://www.nuh.com.sg/personal-data-protection-act>] for more information.

#### **8. Alternatives to genetic testing**

Genetic testing is voluntary, you may choose not to proceed. You may also withdraw from the genetic test process at any point before the test is completed. If consent is withdrawn, the sample will be discarded, and no report will be issued. However, charges would apply once the test request has been received and processed.

## **PART II – DECLARATION AND CONSENT/ ACKNOWLEDGEMENT**

Language used by doctor: \_\_\_\_\_  
Authorised by Doctor? Yes / NA

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**Consent for Treatment/ Operation/ Procedure**

1. I acknowledge that I have been given information on, and understand the nature, purpose, process, risks and alternatives with regard to the treatment/ operation/ procedure listed in **PART I**.
2. I have had the opportunity to discuss and ask for more information about (i) the complications; (ii) the risks in general; and (iii) specific concerns of relevance to me.
3. I consent to undergo the treatment/ operation/ procedure.
4. Secondary or Incidental findings (Applicable to Whole Exome Sequencing/Whole Genome Sequencing only)
  - ☐ I consent to the reporting of secondary or incidental findings.
  - ☐ I do not consent to the reporting of secondary or incidental findings.
5. I acknowledge that my test result(s) and related information may be useful for my family members for their genetic counselling and testing .
  - ☐ I consent to my test result(s) and related information being shared with my family members and the healthcare professionals involved in their care.
  - ☐ I do not consent to my test result(s) and related information being shared with my family members and the healthcare professionals involved in their care.
6. In the event I am uncontactable, the test results may be made known to:  
 Name: \_\_\_\_\_  
 NRIC (last 4 digits): \_\_\_\_\_  
 Contact details: \_\_\_\_\_  
 Relationship: \_\_\_\_\_

**Consent by Donee/ Court-appointed Deputy**

1. If I am the Donee/ Court-appointed Deputy, I confirm that I have the valid authority to consent or refuse consent to carrying out of medical treatment on behalf of the patient.

Y    N    N/A  
☐   ☐   ☐

**Additional Notes**

\_\_\_\_\_  
Name & NRIC of  
Patient/ Parent/ Guardian/ Donee/ Deputy\*  
Consents/ Acknowledges\*

\_\_\_\_\_  
Signature/ Thumbprint (\*Right/ Left) & Date

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### PART III – DECLARATION BY DOCTOR/GENETIC COUNSELLOR

- |                                                                                                                                                                                                                                                                                     | Y                        | N/A                      |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------|--------------------------|
| 1. I confirm that I have explained to the <b>patient/ consenting person</b> the patient's medical condition as well as the nature, purpose, process, risks, complications and alternatives with regard to the treatment/ operation/ procedure and have also addressed their queries | <input type="checkbox"/> | <input type="checkbox"/> |
| 2. I confirm that I have explained to the <b>acknowledging person(s)</b> the patient's medical condition as well as the nature, purpose, process, risks, complications and alternatives with regard to the treatment/ operation/ procedure and have also addressed their queries    | <input type="checkbox"/> | <input type="checkbox"/> |
| 3. I confirm that the proposed treatment/ operation/ procedure is in the <b>best interests of the patient</b> .                                                                                                                                                                     |                          |                          |

### PART IV – DECLARATION BY TRANSLATOR (if applicable)

1. I confirm that I have translated the communication of the doctor named above to the patient/ consenting/ acknowledging person, in \_\_\_\_\_ language/ dialect.

\_\_\_\_\_  
Name & last 4-digits of NRIC/Employee No of  
Translator

\_\_\_\_\_  
Signature of Translator & Date