Centre for Cellular and Molecular Biology (CSIR-CCMB)

Medical Biotechnology Complex, CCMB Annexe II,
Genpact Road, Habsiguda, Hyderabad – 500039

Patient Consent for Next Generation Sequencing Test

Patient Name: _____

Address: _____

_____

Age/DOB: _____ Sex: _____

Dear patient,

Your healthcare provider has recommended a genetic analysis for you (or a person in your legal custody) to clarify the diagnosis/symptoms stated below. In order to ensure that you have understood the purpose and significance of a genetic analysis, we have provided information about the testing process and potential results below.

Please put a tick mark [√] to each statement as acknowledgment that you have read and understand the information.

1. *I understand the following information regarding the test purpose and methodology*: ☐

The purpose of this molecular genetic test is to ascertain the cause of a suspected disease in you or your family by analyzing your genetic material (DNA/RNA) for an abnormal change (variant) that could explain the disease affecting you or the members of your family. In addition with testing of a single gene/variant responsible for a specific, suspected genetic disease, you have the option of testing multiple genes in parallel using Next Generation Sequencing (NGS) methodologies (gene panels, exome, genome or transcriptome sequencing).

The blood, body fluid, or tissue specimen collected/submitted is required for isolation and purification of DNA and RNA (nucleic acids) for molecular genetic testing.

2. *I understand the following information regarding CCMB results disclosure policy*: ☐

Due to the complexity of nucleic acid-based testing and the important implications of the test results, these results will be reported through your designated healthcare providers and their medical team or genetic counselor and you should contact your provider to obtain the results of the test. Additionally, the test results could be released to all who, by law, may have access to such data. Genetic information found from this test will remain in your medical record.

3. *I understand the following information regarding test results*: ☐

Genes included on this test may be associated with several different types of the phenotype (clinical features) and are also may be associated with varying levels of risk. A genetic analysis can have one of several outcomes:

- A disease-causing variant is identified confirming the diagnosis and allowing appropriate medical management by your healthcare provider (if such is available).
- A variant is identified but at this time, there is not enough scientific and medical evidence to determine if this is a disease-causing variant or not. Your healthcare provider will discuss such a result with you and explain what further options are available to you.
- The genetic analysis results in no specific finding that can explain the clinical features. This can be due to the current limitations in scientific or medical knowledge and technology.

It is important to understand that genetic analyses – even if the result of a specific analysis is negative - are not exhaustive and that it is therefore not possible to exclude risks for all possible genetic diseases for yourself and your family members (especially children).

It is possible that the knowledge of the test results may cause psychological stress to you and your family. It is always recommended to discuss the results with your responsible healthcare provider.

Pre- and post-test genetic counseling provided by a qualified specialist, such as a genetic counselor or medical geneticist, is a recommended option for all individuals undergoing genetic testing.

4. *I understand that this molecular genetic test may require an additional blood, body fluid, or tissue sample to obtain accurate results*. ☐

5. *I understand the policy of CCMB for reporting Incidental/secondary findings*: ☐

Genetic analyses, particularly those involving a large number of genes such as exome or genome sequencing, may identify results that are not directly related to the actual reason for your testing (incidental/secondary findings). However, such findings could still be of medical importance for you and your family, as they may provide information about a risk (that you may not be aware of) for potentially serious, unavoidable or non-treatable genetic diseases.

Such findings will be provided only upon your request. Indicate your choice here. YES ☐ NO ☐

6. *I understand the following information regarding technical limitations of this testing and the possibility for additional testing*: ☐
While this test is designed to identify most detectable variations in the genes analyzed, it is still possible that there are variations that this testing technology is unable to detect. In addition, there may be other genes associated with the phenotype that are not included in this analysis or that are not known at this time. Due to the complexity of genetic alterations, the results of the sequencing may not be clear or may require further testing at a later date to confirm or understand the genetic changes. Due to updates in medical knowledge, your healthcare provider may wish to order a reanalysis of your prior test or a new test. If so, a new, updated or amended report will be issued to the ordering healthcare provider, unless you decide to opt out of this.

7. *I understand the following information regarding standard laboratory limitations*: ☐
Imprecise results may occur as a result of (but not limited to) the following reasons: samples unavailable from critical family members, inaccurate reporting of family relationships, inaccurate or misleading medical information about your clinical condition or that of your family members, or technical problems.

8. *I understand the following information regarding use of specimens for research*: ☐
After testing is completed, your blood, body fluid, tissue specimen(s), or DNA sequence may be disposed of or retained indefinitely for medical research, test validation, publication, and/or education by CCMB, as long as your privacy is maintained, without further written consent from you. No compensation will be given nor will funds be forthcoming due to any invention(s) resulting from research and development using the specimens submitted. You may refuse to submit your specimen for use in this way and may withdraw your consent at any time by contacting the laboratory head/director. Your refusal to consent to medical research will not affect your results. Indicate your denial below.

I do NOT consent to the use of my sample for research purposes. ☐

9. *I understand that the genetic information collected from me and my family may be published in scientific journals worldwide for the benefit of the society however the identity of my family will be kept strictly confidential*. ☐

10. *I understand that CCMB reserves the right to*: ☐

- Suggest additional molecular testing if it would help in resolving your diagnosis.
- Refuse testing if one of the conditions in this informed consent document is not met.

11. *I understand the following information regarding my financial responsibility*: ☐
If the test is cancelled before any processing, there will be no charge. If the test is cancelled after nucleic acid isolation and sequencing library construction but before sequence analysis, the patient will be charged the cost associated with the completed technical work and testing will not be done. If the test is cancelled after the sequencing has begun, the patient will be charged the full cost of the test.

GENETIC ANALYSIS FOR DISORDER (to be filled by healthcare provider): _____

I have read or have had read to me all of the above statements and understand the information regarding molecular genetics testing and have had the opportunity to ask questions I might have about the testing, the procedure, the risks, and the alternatives prior to my informed consent. I agree to have the molecular genetic testing.

By signing below, I hereby authorize Dr. _____ to obtain _____ sample from _____

_____ of _____ for the genetic analysis of _____

_____

_____	_____
Name of Patient/Parent/Legal Guardian	Patient/Parent/Legal Guardian's signature & date
_____	_____
Name of person who explained consent	Signature & date
_____	_____
Witness' name (if the parent/legal guardian does not read English)	Witness' signature & date

If you have any questions about this test, please contact at **040-27195612**.