

## INFORMED CONSENT FORM FOR GENETIC TESTING

### What is genetic testing?

DNA provides the information necessary for body's growth and development. Genetic disorders are caused by changes in the DNA and genetic tests identify changes in chromosomes and the DNA of a gene. The purpose of this test is to see if I, or my child/foetus, have a genetic variant or chromosome rearrangement causing a genetic disorder. Additional information about the specific test being ordered is available from my health care provider. This information includes the specific types of genetic disorders that can be identified by the genetic test, the likelihood of a positive result, and the limitations of genetic testing. Genetic testing utilizes many techniques like PCR, Sanger sequencing or Next Generation Sequencing (NGS).

**Information provided by the genetic test:** If I/my child have a family history of one of the conditions that is being tested and is known, then the information must be provided to the lab. A negative result indicates that no disease-causing genetic variant was identified for the test performed. It does not guarantee that I/my child will be healthy or free from other genetic disorders or medical conditions. Result interpretation is based on currently available information in literature and scientific databases. Scientific knowledge is constantly updated, new information that becomes available in the future may replace or add to the information used to interpret my/my child's results. Reanalysis upon formal request will be done by BioServe Biotechnologies (India) Private Limited (BBIPL) and may be charged in some situations. Tests like Clinical exome and Whole exome often reveal additional information than what was being originally investigated. Such findings are called Incidental findings or Secondary findings. Such findings when actionable will be reported by BBIPL.

**Limitations of genetic test:** Though genetic testing is an important part of the diagnostic process; genetic tests may not always give a definitive answer. In some cases, testing may not identify a genetic variant even though one exists. This may be due to limitations in current medical knowledge or testing technology. Test results are interpreted in the context of clinical findings, family history and other laboratory data. Only variations in genes potentially related to the proband's medical condition are reported. Due to inherent technology limitations of the assay, not all bases of the exome/NGS panel can be covered by this test. Accordingly, variants in regions of insufficient coverage or for Genes with pseudogenes and paralog genes are expected to decrease sensitivity and specificity of variant detection and interpretation. Pathogenic variants may be present in this region and are not covered by this test and therefore are not reported. Coverage of the exome/NGS panel genes will be provided upon request.

BBIPL recommends genetic counselling before and after having the genetic test. The pre-test counselling is believed to have covered results to be expected, the chance of finding a clinically significant result, TAT, possibility of no- result, inclusion or exclusion of incidental findings disclosure. The possibility of storing data and use of de-identified data for research purposed.

**Patient Consent:** I have read the Informed Consent document and I give permission to BioServe Biotechnologies (India) Private Limited to perform genetic testing as described on my biological sample. I understand any possible benefits /risks involved in such testing. I also give permission for my specimen / genetic data to be used in (de-identified) studies at BBIPL to improve genetic testing for other patients. In case of prenatal specimen, the sex of my fetus will not be revealed, neither shall I seek such information from BBIPL.

### Address

Pinnacle Towers, 1st Floor, Plot No.9/17/A&B Road  
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Name & Signature

Date