PATIENT INFORMED CONSENT AND INSURANCE ACKNOWLEDGEMENT

GENETIC CARRIER SCREENING

Important – Please Read Carefully: This Patient Informed Consent and Insurance Acknowledgement issued by Medical Diagnostic Laboratories ("MDL") describes the purpose, procedure, benefits, limitations and possible risks of genetic carrier screening. This is voluntary screening; therefore, you may wish to seek genetic counseling prior to signing this form.

Purpose: The primary purpose of genetic carrier screening is to identify carrier status in disease genes and the risk for passing on genetic conditions to offspring. Genetic testing is an increasingly important part of family planning. Usually, carriers are healthy individuals with no symptoms; however, when two parents are carriers of the same genetic disease this can result in an affected child. By analyzing DNA, your risk of being a "carrier" of a recessive genetic disease can be estimated. Using this and other information, your overall risk (aka "reproductive risk") for having a child with a genetic disorder is calculated. This risk is best assessed as a paired analysis with consideration of both parental ethnicities.

Screening Procedure: This screening requires Genomic DNA extracted from a blood, saliva, or ***OneSwab***® sample. High-throughput next generation sequencing is performed to examine over 1,300 DNA variants associated with multiple diseases. In some instances, an additional sample may be needed if the volume, quality and/or condition of the initial specimen is not adequate for testing.

No additional screening will be performed, without specific, signed authorization. After 60 days, unless consent is given, the sample will be destroyed or will be de-identified from all patient information and used for standard laboratory purposes, as a positive or negative control for this screening.

Screening Results: MDL's variant classification system is based on the 5-tier system recommendations for the interpretation of sequence variants proposed by the American College of Medical Genetics and Genomics (ACMG) and complies with the standards and guidelines for the interpretation of sequence variants by ACMG and the Association for Molecular Pathology (AMP). Screening results are reported using numbering and nomenclature recommended by the Human Genome Variation Society (HGVS http://hgvs.org). All results are reported in reference to Human Genome 19, Human Build 37.5**.**

- Any detected variants that are a recognized cause of the disease (Pathogenic) will be reported.
- In addition, variants that have not previously been established as a recognized cause of disease may be identified. In these cases, only variants classified as "likely" pathogenic are reported.
- Benign variants, likely benign variants and variants of uncertain significance, and variants not directly associated with the intended disease phenotype are not reported.

The screening results become a part of your medical record, and may be made available to individuals and/or organizations with legal access to your medical record, on a strict "need-to-know" basis including, but not limited to the physicians and nursing staff directly involved in your care, your genetic counselor, current and future insurance carriers and others specifically authorized by you or an authorized representative to gain access to your medical records. MDL maintains the confidentiality of your tests results in full compliance with the Health Insurance Portability and Accountability Act (HIPAA) and applicable state laws.

Screening Interpretation: Your physician and/or genetic counselor will evaluate and discuss with you the results of your genetic carrier screening taking into consideration your personal and family history, clinical information and laboratory data.

The Federal Genetic Information Nondiscrimination Act of 2008 (GINA) protects individuals from any type of discrimination based on the results of genetic testing. For additional information about GINA and the state laws that also protect against discrimination based on the results of genetic testing, visit www.ginahelp.org.

Because the results of genetic screening have implications for your blood relatives, in consultation with your physician or genetic counselor, you may wish to discuss sharing your test results with certain blood relatives who may be at-risk. Genetic carrier screening may provide previously unknown information about relationships in families.

Screening Benefits: The results of genetic carrier screening will assist you and your physician in making more informed choices relevant to your medical management, health care options and treatment. MDL's expanded carrier screening panel provides not only currently recommended carrier screening tests, but also information pertaining to other high-risk inherited genetic conditions. (**Table 1** contains a complete list of detected diseases, targeted genes and frequencies.) Identification of a pathogenic variant in one of these genes, or variants in genes associated with high-risk diseases, can help health care providers or genetic counselors establish or confirm a diagnosis, predict the risk of having a child with a genetic disorder, or guide patients' managing decisions.

Screening Risks: This screening requires a saliva or blood sample. Side effects from the venipuncture may include pallor, sweating, nausea, dizziness, light-headedness and fainting. Also there may be pain, swelling or bruising around the site where the blood was drawn.

Screening Limitations: This test determines the presence of variants in select gene(s). (**Table 1** provides a list of the select genes tested for the presence of variants.) This test is not the only way to detect genetic abnormalities. Your health care provider may recommend other genetic, imaging or laboratory tests.

Residual Risk: Residual Risk is the possibility of being a carrier in the case of a negative test result for any genetic disease tested. A negative result for any of the diseases tested in the MDL carrier screening panel significantly decreases a person's risk of being a carrier for this genetic condition. (**Table 1** contains the Residual Risk estimates for the carrier conditions in the MDL genetic carrier screening test.) However, it is still possible to be a carrier for a disease-associated variant that is not tested utilizing the MDL genetic carrier screening test.

Patient Statement of Informed Consent

By signing below, I have read and fully understand this form and acknowledge the following:

1. I have been informed by my physician and/or genetic counselor of the purpose, procedure, benefits, limitations and possible risks of this genetic test. I have been given the opportunity to ask and have all my questions answered about this genetic test.

2. I have discussed with my physician and/or genetic counselor ordering this test, the reliability of the positive or negative test results and the level of certainty that a positive test result for a gene variant serves as a predictor of passing on a genetic condition to an offspring.

3. I have read this entire document and have been informed that I may retain a copy for my records.

4. I will discuss the screening results and appropriate medical management with my physician and/or genetic counselor.

I have read and fully understand the above, and give my consent to the performance of this **genetic carrier screening test** and accept the consequences of this decision.

Patient Name: _____ **Patient/Legal Guardian Signature**:

Date:

Patient Insurance Acknowledgement

Genetic carrier screening testing determined to be medically necessary by your physician or genetic counselor is usually reimbursed by health insurance. However, Medicare, Medicaid and some insurance carriers may not pay for this test. Certain insurance plans in order to receive prior authorization to perform the test, require the patient to participate in pre-test genetic counseling.

By signing below, I have read and fully understand, and acknowledge the following:

1. I further understand that I am financially responsible for any amounts not covered by my insurer for this genetic test. MDL will perform the genetic test and bill me without further contact if my total financial responsibility will not exceed $149.00 for any reason, including co-insurance, co-payments, deductibles or non-covered services. If the out-of-pocket expense will exceed $149.00, I will be contacted to discuss my financial responsibility.

2. I authorize MDL to furnish my designated insurance carrier the information on this form, if necessary for reimbursement. I also authorize MDL to release medical information concerning genetic carrier screening to my insurer and, if applicable, I authorize MDL to be my Designated Representative for purposes of appealing a denial of benefits.

3. For direct insurance/third-party billing, I hereby authorize my insurance benefits to be paid directly to MDL.

4. I understand that I am legally responsible for sending MDL any money received from my health insurance company for the performance of this genetic test.

5. I understand that Medicare/Insurance Carriers may not pay for genetic carrier screening. I also understand that if I am a patient with Medicare insurance, I may be required to complete an Advance Beneficiary Notice.

6. I understand that my medical history and these genetic carrier screening results will not be discussed or disclosed to a third-party, unless related to my treatment or payment for my treatment, without my express written authorization.

7. I understand that MDL will honor test screening cancellations received prior to the initiation of the test. Once the genetic screening testing has been initiated, no cancellation request will be honored and a claim will be submitted to your insurance company and the test results will be provided to the ordering physician.

I fully **understand and agree to my financial responsibilities** concerning the performance of this genetic test.

Patient Name: _____ **Patient/Legal Guardian Signature:**

Date:

Table 1. Detected Diseases, Target Genes and Frequencies.

Disease	Gene	Population	Carrier Frequency	Detection Rate	Residual Risk
Cystic Fibrosis	**CFTR**	Caucasian	1 in 25	94%	1 in 417
		Asian	1 in 94	65%	1 in 269
		Ashkenazi	1 in 24	97%	1 in 800
		General	<1 in 250	95%	1 in 5,000
		General	<1 in 500	95%	1 in 10,000
Spinal Muscular Atrophy	**SMN1**	Ashkenazi	1 in 41	94%	1 in 683
		European Caucasian	1 in 354	95%	1 in 700
		Asian	1 in 53	93%	1 in 757
		Hispanic	1 in 117	95%	1 in 2,340
		General	1 in 500	10%	1 in 556
		Finnish	1 in 70	10%	1 in 78
		French Canadian	1 in 55	95%	1 in 21,620