

## Patient Informed Consent for Genetic Testing

### Test Specific Information (check those that apply)

**MM490 Spinal Muscular Atrophy: Carrier Screen**

Spinal muscular atrophy (SMA) is the second most common, autosomal recessive disorder in Caucasians, with a carrier frequency of 1/50. SMA is a common cause of infant death and is characterized by severe and progressive symmetrical muscle weakness and wasting, joint contractures, respiratory insufficiency, and feeding and sleep difficulties. It has variable age of onset and severity of symptoms.

Limitations:

- This carrier assay tests for the common SMN1 deletion (exon 7 and 8 deletion) only; single point mutations or variants other than SMN1 gene will not be detected.
- Approximately 5-8% of carrier individuals will have a deletion on one chromosome with two normal *SMN1* copies on the second chromosome. This assay will not detect these carrier individuals.
- This assay will not report *SMN2* deletions.

**MFRAX Fragile X: CGG Repeat Analysis**

Fragile X syndrome is the most common inherited (X-linked) form of intellectual disability. Additional symptoms include autism, behavioral problems, and characteristic facial features in affected males. This test can also identify premutation carrier females who are at risk for developing premature ovarian insufficiency characterized by infertility, early menopause, and other ovarian problems, and premutation carrier males who are at risk for developing late onset fragile X-associated tremor/ataxia syndrome characterized by problems with movement and thinking ability after approximately the age of 50.

Limitation:

- Approximately 1% of cases of *FMR1*-associated intellectual disability are due to mutations that cannot be detected by this test. Other testing such as sequencing and deletion/duplication of the *FMR1* gene might be warranted.

**CF Cystic Fibrosis: *CFTR* Common Mutation Panel [Dried blood sample submission only]**

**MM530 Cystic Fibrosis: *CFTR* Expanded Mutation Panel [Blood sample submission only]**

*CFTR* Common Mutation Panel tests for only the 39 most common mutations found in a Caucasian population, while the expanded panel tests for 142 mutations that are found across all ethnic groups. Cystic fibrosis (CF) is an autosomal recessive disorder involving multiple organ systems. Classic CF primarily involves the respiratory and digestive systems, and may have a range of clinical severity. Thick mucus accumulation in the lungs leads to breathing difficulty, infection and poor digestion. The average life expectancy is in the 30s. Congenital bilateral absence of the vas deferens (CBAVD) is seen in men without pulmonary or digestive symptoms of CF, and results in absence of sperm in the semen.

Limitations: Pathogenic variants in regions other than the targeted area will not be detected by this analysis.

**MM580 ACOG/ACMG Carrier Screen: Targeted Mutation Panel**

- The ACOG/ACMG Carrier Screen tests for Cystic Fibrosis, Spinal Muscular Atrophy, and eight other disorders common to those of Ashkenazi Jewish descent. This screen is limited to those conditions recommended by the American College of Obstetrics and Gynecology and the American College of Medical Genetics for screening during pregnancy.
- Fragile X testing is not currently recommended by ACOG/ACMG unless there is a family history and therefore not included in this panel. Fragile X testing may be ordered separately.

Limitations: Pathogenic variants in regions other than the targeted area will not be detected by this analysis.

**MM480 Pan-Ethnic Carrier Screen: Targeted Mutation Panel**

- The Pan-Ethnic Carrier Screen tests for 148 genes that cause autosomal recessive and X-linked conditions. Includes Cystic Fibrosis, Spinal Muscular Atrophy, Fragile X syndrome, and 143 other genetic disorders related to intellectual disability, mobility impairment, visual impairment, joint and bone disorders, nervous system abnormalities, developmental delay, hearing loss, skin irregularities, and metabolic syndromes.
- Includes only most common mutations in each disorder.

Limitations: Pathogenic variants in regions other than the targeted area will not be detected by this analysis.

**MM470 Pan-Ethnic Carrier Screen: Gene Sequencing Panel**

- The Pan-Ethnic Carrier Screen tests for 148 genes that cause autosomal recessive and X-linked conditions. Includes Cystic Fibrosis, Spinal Muscular Atrophy, Fragile X syndrome, and 143 other genetic disorders related to intellectual disability, mobility impairment, visual impairment, joint and bone disorders, nervous system abnormalities, developmental delay, hearing loss, skin irregularities, and metabolic syndromes.
- Looks at entire coding region for more mutations and rarer mutations.

Limitation:

- Due to technical issues the results may be inconclusive and the test might need repeating. Results may also be inconclusive due to the identification of a variant of unknown significance.



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### General Information, Limitations, and Risks:

- This test is indicated for individuals or couples seeking to assess reproductive risk for a variety of conditions and individuals or couples that are considered high risk due to ethnicity, family history, or clinical indication.
- Carriers of autosomal recessive disorders are typically healthy individuals but can pass on their genetic variants to children. If both partners are identified as carriers of the same genetic disorder, they have a 25% chance of having a child affected with that disorder with each pregnancy.
- Women who are carriers of X-linked conditions are typically healthy individuals but have a 50% chance of having a son who is affected with the condition or a 50% chance of having a daughter who is also a carrier. Carrier can sometimes be mildly affected.
- DNA testing requires a blood sample or saliva sample, both of which have risks associated with obtaining the sample. Additional samples may be needed if the sample is damaged in shipment or inaccurately submitted. In order to perform accurate prenatal testing, samples from the affected individual, parents, or additional family members may be required.
- DNA-based studies performed are specific to the condition(s) indicated above. The accuracy of genetic testing is limited by the methods employed, the clinical diagnosis, and the nature of the specific condition for which testing is requested. In some cases, the test will detect an abnormality, called a mutation, in the gene. In other cases the test is unable to identify an abnormality although an abnormality may still exist. This event may be due to the current lack of knowledge of the complete gene structure or an inability of the current technology to identify certain types of changes (mutations) in a gene.
- As with any complex genetic test, there is always a small possibility of a failure or error in sample analysis. Extensive measures are taken to try to avoid these errors. The methods are not 100% accurate due to the possibility of rare genetic variations in the DNA of an individual or due to the complexity of the testing itself. A low error rate, approximately 1 in 1000 samples, is generally estimated to exist in a laboratory.
- Due to the complexity of DNA testing and potential implications of test results, results will be reported directly to the patient's ordering provider, who will then review and discuss the test results with me.
- Patient-identifying results and information at Eurofins NTD, LLC will remain confidential and may only be released to other parties with my expressed written consent or as permitted or required by applicable law.
- I understand no tests other than those authorized shall be performed on my sample and that the sample shall be discarded within sixty days after taken. However Eurofins NTD, LLC performs research and development studies to improve and to validate existing and new tests and to advance biomedical knowledge. I and my heirs will not receive payments, benefits, or rights to any resulting products or discoveries. Patient permission is requested for the use of patient de-identified sample in research and development studies and is entirely voluntary.
- If I have additional questions, I understand that I may wish to obtain further professional genetic counseling prior to consenting to this testing.

My signature below acknowledges my voluntary participation in the genetic tests ordered by my physician and verifies that I have been appropriately counseled about the testing process and the different possible outcomes.

\_\_\_\_\_  
Patient/Guardian Signature

\_\_\_\_\_  
Printed Name

\_\_\_\_\_  
Date

### Physician/Counselor/Clinician Statement:

I have explained DNA testing to the patient/parent/guardian. The consent form and limitations of genetic testing were reviewed with the patient/parent/guardian. I accept responsibility for pre- and post-test genetic counseling. I will use my independent professional judgment and the patient's best interests in advising the patient/parent/guardian regarding DNA test results, the use and limitations of same, and any research study, clinical trial, drug, treatment or device brought to my attention by Eurofins NTD, LLC or others.

\_\_\_\_\_  
Healthcare Provider Signature

\_\_\_\_\_  
Printed Name

\_\_\_\_\_  
Date

**A signed physician acknowledgement of NY informed consent for genetic testing must be on file at Eurofins NTD, LLC to permit testing and processing.**

**This Patient Informed Consent form is to be maintained by the physician in the patients' medical file. DO NOT Return to Eurofins, NTD, LLC.**