Laboratory of Personalized Genomic Medicine
Department of Pathology and Cell Biology

COLUMBIA UNIVERSITY MEDICAL CENTER

630 West 168th Street
P&S 11th Floor, Room 453
New York, NY 10032
Tel: 212-305-9706
Fax: 212-342-0420
PGMINQUIRY@cumc.columbia.edu

Informed Consent– *C9orf72* Repeat Expansion Testing

Please read the following carefully and discuss with your ordering physician/genetic counselor before signing consent.

1. The purpose of this analysis is to detect pathogenic mutations in your DNA associated with Amyotrophic Lateral Sclerosis/Frontotemporal Degeneration (ALS/FTD.)

    1a. You (or the person undergoing testing) may request genetic counseling before signing consent.

2. The condition being tested is ALS/FTD, caused by expansion of the GGGGCC repeat in the gene *C9orf72*. This expansion is the most common cause of ALS/FTD.

3. A positive finding indicates that the patient's ALS/FTD is due to expansion in the *C9orf72* gene. Since this expansion is heritable, other members of your family might carry it and have a risk to develop ALS/FTD. If the test is positive for the disorder, you may wish to have further independent testing, consult your physician or have professional genetic counseling.

5. This is a test for determining the cause of ongoing disease. It is not capable of predicting with certainty that a non-symptomatic person carrying the mutation will develop the disease during his/her lifetime. The risk of having the disorder may be altered by family history and/or other factors.

6. A positive test indicates that the patients ALS/FTD is due to expansion in the C9orf72 gene. A negative result indicates that the disease is caused by another mutation previously described or unknown. Consult a genetic counselor to discuss if additional testing is appropriate.

7. The results of the above test become a part of the patient's medical record, and may be made available to Individuals/organizations with legal access to the patient's medical record, on a strict "need-to-know" basis, including, but not limited to the physicians and nursing staff directly involved in the patient's care, the patient's current and future insurance carriers, and others specifically authorized by the patient authorized representative to gain access to the patient's medical records. Columbia University, NewYork-Presbyterian and Weill Cornell Medicine and their related entities participate in an Organized Health Care Arrangement (OHCA). This allows us to share health information to carry out treatment, payment and our joint health care operations, including integrated information system management, health information exchange, financial and billing services, insurance services, insurance, quality improvement, and risk management activities. Organizations that will follow this Notice include Columbia University, NewYork-Presbyterian sites, Weill Cornell Medicine and their related entities.

8. No additional tests will be performed on this sample, without specific, signed authorization by the patient. After 60 days, unless consent is given the sample will be destroyed- please see below.

9. Medicare/Insurance Carriers may not pay for the test, in which case, the patient/responsible party will be billed for the test.

Name of Person Obtaining Consent:

_____ _____ **Date:** _____

Print name of Person Obtaining Consent **Signature of Person Obtaining Consent**

_____ _____ **Date:** _____

Print Name of Patient/Authorized Representative **Signature of Patient/Authorized Representative**

Relationship to Patient (if parent or legal guardian): _____

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Consent for Sample Retention:

I consent to the retention of this sample for: (check and sign on appropriate line)

☐ I do not consent to research. My sample may be used for routine laboratory use only.

_____

Signature

☐ I consent to possible future genetic research on my specimen, only if all identifying information is removed (name, address, date of birth, medical record number). Since my identifying information will be removed, I will not be contacted with any research results. The duration of the retention of my sample will depend on the individual research study. If the sample is not used in a study, it will be destroyed or anonymously used as described above.

_____

Signature