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- Huntingtin (HTT) CAG Repeat Expansion Testing

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

1. This is a test for expansion of a CAG repeat in the HTT (Huntingtin) gene, by PCR.
2. The purpose of this analysis is to confirm a diagnosis of Huntington's Disease (HD), or to evaluate risk for development of HD.
   2a. Genetic counseling is strongly recommended before you sign the consent.
3. This is a test for genetic susceptibility ("genetic predisposition"), the risk of having the disorder may be altered by family history and/or other factors. If the test is positive for the disorder or for an increased risk of the disorder, the patient may wish to have further independent testing, consult your physician or have further genetic counseling.
4. The condition being tested is Huntington's disease, a progressive, fatal, neurological disorder.
5. If the result is "normal", then the patient will not be at increased risk for having HD. If the test shows an "HD allele" the patient will develop HD at some point in his/her life; if the result shows a "mutable normal allele" then the patient will not be at increased risk for HD, but his/her descendants may be at increased risk for developing HD. If the result of the test is "HD allele with reduced penetrance", then the patient may or may not develop HD during your lifetime, although the risk with be higher than the general population.
6. 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.
7. No additional tests will be performed on this sample, without specific, signed authorization by the patient/authorized representative. After 60 days, unless consent is given the sample will be destroyed, or will be de-linked from all patient information and used for standard laboratory quality assurance purposes.
8. 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**

I have read and fully understood the above and give my consent for this testing.
Patient (person being tested):

_____ _____ **Date:** _____

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

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

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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