Penn Medicine

To: UPHS Physicians and Staff
From: Department of Pathology and Laboratory Medicine
Division of Precision and Computational Diagnostics, Molecular Pathology Laboratory
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Date: November 18, 2019
Re: NEW ASSAY: *HTT* CAG Repeat Expansion Analysis for Huntington Disease

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In November, CAG repeat expansion testing for the *huntingtin* gene (*HTT)* will be available in the Molecular Pathology Laboratory at the Hospital of the University of Pennsylvania for diagnostic, predictive and prodromal genetic testing for Huntington Disease, a neurodegenerative disease characterized by progressive motor, cognitive, and neuropsychiatric dysfunction. Huntington disease is caused by an expansion in the number of CAG repeats in exon 1 of *HTT*. The differential diagnosis for Huntington disease includes frontotemporal degeneration and amyotrophic lateral sclerosis. This PCR-based assay can detect and size alleles of up to 200 CAG repeats. Larger may be detected, but not accurately sized.

Result Reporting Categories

≤26 CAG repeats	Normal
27-35 CAG repeats	Intermediate allele
36-39 CAG repeats	Reduced penetrance allele
≥40 CAG repeats	Expansion associated with fully penetrant Huntington disease

Testing Information

Turnaround time: Up to 15 business days

Ordering: Huntington Disease (PxCode HTTCASE) in PennChart

Acceptable specimens:

Peripheral blood collected in an EDTA (lavender) tube
OraCollect-Dx oral swabs (Lawson # 281926)
Frozen tissue (call the laboratory to discuss)

Informed Consent: A consent form, signed by the patient and the ordering physician/genetic counselor must be kept on file by the ordering physician/genetic counselor.

Contact Information: Call the Molecular Pathology Laboratory (215-662-6121) weekdays during regular business hours. For information on ordering and specimen requirements, kindly consult the Lab Tests Services Guide.