

Huntington's Disease

Order Name: **HUNTINGTON**

Test Number: 5562725

Revision Date: 02/13/2023

| TEST NAME | METHODOLOGY | LOINC CODE |
|----------------------|----------------------------|------------|
| Huntington's Disease | Repeat-Primed PCR (QP-PCR) | |

| SPECIMEN REQUIREMENTS | | | | |
|-----------------------|---|------------------|---------------------|-----------------------|
| Specimen | Specimen Volume (min) | Specimen Type | Specimen Container | Transport Environment |
| Preferred | 5 mL (3 mL) | EDTA Whole Blood | EDTA (Lavender Top) | Room Temperature |
| Instructions | For children/adults: prefer 2 lavender top tubes For newborns: prefer 1-2 mL Click here for Presymptomatic Huntington Disease Testing form. This consent form is required for testing to be performed by the Genetic Center. | | | |

| GENERAL INFORMATION | |
|------------------------------|--|
| Testing Schedule | Varies |
| Expected TAT | 10 Days |
| Clinical Use | Huntington disease (HD) is a neurodegenerative disease of mid-life onset that produces choreic movements and cognitive decline, often accompanied by psychiatric changes. The disease is caused by an expansion of the CAG repeats in 3-5 out of 100,000 individuals. However, the prevalence of HD exceeds 15 per 100,000 in some populations, mostly of Western European origin. Juvenile-onset HD occurs in approximately 5% of affected patients, is rapidly progressive, and presents with rigidity, spasticity, and intellectual decline before the age of 20 years. The symptoms result from the selective loss of neurons, most notably in the caudate nucleus and putamen, and there is currently no effective treatment. |
| Performing Labcorp Test Code | 620016 |
| Notes | Reference Lab: St. Francis St. Francis Test Code: 9584 |
| CPT Code(s) | 81271 |
| Lab Section | Reference Lab |