MAYO CLINIC LABORATORIES

TEST CHANGE

Notification Date: September 10, 2024
Effective Date: October 10, 2024

Huntington Disease, Molecular Analysis, Varies

Test ID: HAD

Explanation: On the effective date, this assay will change the acceptable anticoagulants for the whole blood specimen type. They will now be limited to the anticoagulants shown below. Furthermore, additional specimen and clinical information will be updated/added.

Current Specimen Required	
Specimen Type:	**Whole blood**
Preferred:	Lavender top (EDTA) or yellow top (ACD)
Acceptable:	Any anticoagulant
Specimen Volume:	3 mL
Collection Instructions:	1. Invert several times to mix blood 2. Send whole blood specimen in original tube. **Do not aliquot.**
Minimum Volume:	1 mL

New Specimen Required	
Specimen Type:	**Whole blood**
Required:	Lavender top (EDTA) or yellow top (ACD)
Specimen Volume:	3 mL
Collection Instructions:	1. Invert several times to mix blood 2. Send whole blood specimen in original tube. **Do not aliquot.**
Additional Information:	1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed. 2. To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.
Minimum Volume:	1 mL

Current Clinical Information	New Clinical Information
Huntington disease (HD) is an autosomal dominant progressive neurodegenerative disorder caused by a CAG repeat expansion in the *HTT* gene. HD is associated with cognitive impairment leading to dementia and a wide range of neuropsychiatric problems including apathy, depression, anxiety, and other behavioral disturbances. Additionally, affected individuals typically develop extrapyramidal symptoms (eg, dystonia, dysarthria, chorea, gait disturbance, postural instability, oculomotor dysfunction).	Huntington disease (HD) is an autosomal dominant progressive neurodegenerative disorder associated with progressive involuntary and voluntary motor disturbances (chorea, dystonia, dysarthria, gait disturbance, postural instability, oculomotor dysfunction), cognitive decline leading to dementia, and a wide range of neuropsychiatric problems including apathy, depression, anxiety, and other behavioral disturbances. Onset occurs typically in the late 30's to early 40's, but rare individuals may present with juvenile onset. Huntington disease is caused by a CAG (cystine, adenine, guanine) repeat expansion in the *HTT* gene and is associated with genetic anticipation, whereby repeat sizes may expand with transmission to subsequent generations. Correlation exists between the size of the CAG repeat and disease onset and severity, with larger alleles associated with earlier onset and more severe disease presentation. Full penetrance *HTT* expansions are greater than 39 repeats, while normal alleles are less than 27 repeats. Allele sizes between 36 and 39 repeats are associated with reduced penetrance of clinical HD symptoms. Intermediate alleles (27-35 repeats) are not typically associated with clinical symptoms; however, both reduced penetrance and intermediate alleles may expand into the full penetrance range with transmission to offspring. Identification of a disease-associated repeat expansion has important implications for family members. Testing of at-risk individuals is possible, but it is recommended that predictive testing be performed in conjunction with appropriate pre- and post-test counseling. Additionally, presymptomatic testing of minors is strongly discouraged.

Questions

Contact Michelle Raths, Laboratory Resource Coordinator at 800-533-1710.