

UCLA DIAGNOSTIC MOLECULAR PATHOLOGY LABORATORY
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Huntington Disease

↑CPT

83891; 84311; 83900; 83909; 83912

↑Synonyms

Huntington Chorea

↑Test Includes

Sizing of the CAG trinucleotide repeat expansion in the Huntington gene

↑Laboratory

Molecular Pathology

↑Availability

Monday-Friday, 0700-1700

↑Turnaround Time

3-14 days

↑Specimen

Whole blood

↑Volume

4 mL

↑Container

Lavender top (EDTA) tube

↑ Storage Instructions

All specimens should be sent to the Laboratory immediately after collection, preferably by overnight delivery. Specimens should be kept at room temperature or refrigerated but not frozen.

↑ Causes for Rejection

Blood samples frozen and thawed will yield low quality DNA; specimens inadequately identified

↑ Reference Range

35 CAG trinucleotide repeats or less.

↑ Use

Huntington disease (HD) is an autosomal dominant inherited neurodegenerative disorder which usually presents by the age of 40-50 and is characterized by progressive disordered movements (chorea), psychiatric alterations, and cognitive decline. HD is caused by an expansion of a CAG trinucleotide repeat within a novel gene on 4p16.3. Normal chromosomes usually have 11-35 repeats, while Huntington chromosomes will have larger than or equal to 40, up to 100 or more, repeats. Repeat sizes between 36-39 have variable penetrance. The CAG repeat length shows a highly significant correlation with age of onset of clinical features and this association is particularly evident for persons with juvenile onset HD where CAG expansion is usually >100 repeats.

↑ Limitations

Repeat numbers in the range of 35-39 have been shown to have a variable phenotype and thus should be interpreted in the context of an affected relative.

↑ Methodology

The size of the trinucleotide repeat is determined by performing polymerase chain reaction amplification using fluorescent labeled primers that flank the trinucleotide repeats followed by electrophoresis. The number of repeats can be determined from the peak size by comparison with known size markers. Normal individuals will have allele sizes of 11-28 repeats. Most people will have two different sized bands in this range, but a few will be homozygous for the same size band. Huntington chromosomes will produce bands from 40-120 repeats.

↑Additional Information

Results will be kept confidential and reported only to the referring physician or genetic counselor. Presymptomatic testing is generally offered through the Medical Genetics Clinic and involves stringent informed consent procedures as well as pre- and post-test genetic counseling. These requirements are not mandatory for diagnostic testing in symptomatic individuals.
