

## Huntington Disease (HD) Mutation Testing

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### DESCRIPTION:

- This test detects the genetic abnormality known to cause Huntington disease: CAG trinucleotide repeat expansion in the DNA of the "huntingtin" (IT15) gene located in the chromosomal region 4p16.3. This expansion is present in almost all cases (98-99%) of HD and is absent in controls (Kremer et al., *New Engl J Med*, 1994, 330: 1401). HD symptoms include chorea, discoordination of movement, depression, and memory loss.

### REASONS FOR REFERRAL:

- Confirm or rule out the diagnosis of HD in a clinically symptomatic individual. Please indicate symptoms on DNA referral form.
- As recommended by the HD Society of America, predictive testing of individuals at risk is performed after evaluation and counseling. Shodair Hospital has an approved program (800-447-6614).

### METHOD OF ANALYSIS:

- DNA from the patient is amplified by PCR using fluorescent primers for the CAG region only. Sizes of the CAG repeat alleles are determined by a high resolution laser-induced fluorescence capillary electrophoresis system with internal standard.
- Results are reported within 3 weeks or less of receipt of sample and can be expedited upon request.

### REFERENCE RANGES:

- 26 or fewer CAG repeats are Normal Alleles
- 27-35 CAG repeats are Mutable Normal Alleles (possibly expand in offspring)
- 36 to 39 CAG repeats are HD Alleles with Reduced Penetrance (lead to symptoms in some individuals, but not all, within a "normal" life span. Rubinsztein et al. *Am J Hum Genet*, 1996, 59:16)
- 40 or more CAG repeats are HD Alleles (consistently lead to symptoms of HD)

### SAMPLE REQUIREMENTS:

- For DNA testing, 5 to 10 milliliters of blood (minimum 1 ml) in EDTA (purple top) tubes should be sent by overnight carrier at room temperature.
- Prenatal testing: Please call the laboratory.

### TEST CPT CODES:

- CPT 83891 DNA extraction highly purified
- CPT 83892 DNA enzymatic digestion
- CPT 83894 DNA separation X 2
- CPT 83898 DNA amplification
- CPT 83912 DNA interpretation and report

*Discounts from list price are available for institutional billing under contractual arrangement with the laboratory. Contact Ellen Livers at 800-447-6614 ext 7523.*