Huntington Disease (HD) – OMIM 143100

Background

Huntington disease (or Huntington chorea) is a progressive genetic disorder of the central nervous system affecting muscle co-ordination and some cognitive functions. It is caused by a mutation in the *Huntingtin* (*HTT*) gene (OMIM 613004) and follows an autosomal dominant inheritance pattern, so there is a 50% chance of a child inheriting the mutated gene and having the disease if one parent is affected. Symptoms can appear at any time, from infancy to old age, but usually begins between 35 and 44, and affects both men and women. Individuals can vary in terms of how the disease affects them, even within the same family, but the progression of symptoms follows the same pattern for most people. Early symptoms include a general lack of co-ordination and steadiness, and changes in mood. As the disorder advances, physical co-ordination becomes very difficult and mental abilities decline towards dementia. Complications arising from ever-worsening symptoms mean that life expectancy is usually about twenty years after the symptoms first appear. HD demonstrates anticipation where age of onset decreases and severity increases with inheritance through a multi-generational family.

Recommended Clinical Referral Criteria

- Diagnostic cases and predictive testing for at risk family members
- Autosomal dominant inheritance
- Progressive motor disability, mental disturbances

Molecular Analysis

Mutation screen: Fragment length analysis of *HTT* gene [CAG]n triplet repeat expansions in exon 1 by PCR and triplet primed PCR – the clinical sensitivity of this test is >99% in patients with HD.
Family follow-up: Testing for known familial mutations in the *HTT* gene

Test (Price available on request)	**TAT (calendar days)**
Diagnostic *HTT* gene expansion analysis (R68)	42
Predictive *HTT* gene expansion analysis	28
Linkage analysis for *HTT* (R383)	Performed at an external laboratory.

Contact Details
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Accredited to ISO 15189:2012
(8988)

Sample Requirements
Blood – 5ml in EDTA (1ml neonates/infants);
Please contact lab prior to sending a prenatal sample.
Please label samples with three identifiers and date of collection
All samples must be accompanied by request form
Consent for testing & DNA storage is assumed when request for test received

Links

Orphanet
http://www.orpha.net/
OMIM
http://www.omim.org/
Genetic Test Registry
http://www.ncbi.nlm.nih.gov/gtr/

Support
http://www.hda.org.uk/