INTRODUCTION
HD is a progressive disorder of motor, cognitive, and psychiatric disturbances. Approximately, two thirds of affected individuals present with neurological manifestations, whilst others present with psychiatric changes. Juvenile HD is defined by the onset of symptoms before the age of 21 years.
HD is inherited in an autosomal dominant manner. Differential diagnoses of HD include DRPLA, HDL1, HDL2 and SCA17. The prevalence of Huntington disease is estimated to be between 3 and 7 per 100,000 in populations of Western European descent.

The HTT (or IT15) gene is located on 4p16.3 and is the only gene associated with Huntington disease. An expansion of a trinucleotide (CAG)n repeat in the coding region (exon 1) of HTT is the only mutation observed.

TESTING
Diagnostic:
- For adults and juveniles suspected of having Huntington disease
Presymptomatic:
- Referrals for asymptomatic individuals with clinically affected relatives
Prenatal:
- Prenatal testing must be discussed with the laboratory and arranged in advance
Exclusion:
- Exclusion testing must be discussed with the laboratory and arranged in advance.

REFERRALS
Diagnostic
- Clinical Genetics, Clinical Neurology, Clinical Psychiatry, Paediatric Neurology
Presymptomatic
- Clinical Genetics
Prenatal
- Clinical Genetics in conjunction with Prenatal Diagnosis Department
Exclusion
- Clinical Genetics

TECHNICAL INFORMATION AND STRATEGY
- Fluorescent PCR across the HD (CAG)n repeat for CAG repeat size determination
- PCR across both the HD (CAG)n repeat and nearby (CCG)n repeat for compound repeat size determination
- Triplet primed PCR across the HD (CAG)n repeat
  - Confirms the presence/absence of a (CAG)n expansion only; no sizing information is provided
- Linked marker analysis
  - For exclusion testing
  - Provides a second independent test for prenatal diagnosis, also enables exclusion of maternal contamination

TARGET REPORTING TIMES
Diagnostic test: 10 working days
Presymptomatic test: 10 working days
Prenatal test: 3 working days
Exclusion testing preparation: 40 working days

N.B. Details are correct for the date of printing only – last updated 02/09/2015