Fragile X tremor/ataxia syndrome (OMIM# 300623)

Background

Fragile X tremor/ataxia syndrome (FXTAS) is characterised by late-onset progressive cerebellar ataxia and intention tremor.

FXTAS is caused by a premutation-sized expansion of a CGG repeat within the 5'UTR of the *FMR1* gene (OMIM* 309550). Approximately 1/3 of males with premutations of the *FMR1* gene over the age of 50 are found to have symptoms of FXTAS.

FXTAS has also been identified in females with *FMR1* premutations, although clinical features may be more subtle and occur less often than in male counterparts.

FMR1 expansions in the premutation size range are likely to be unstable and prone to further expansion in the next generation. The identification of a premutation may therefore have implications for other family members who may inherit this expansion.

Recommended Clinical Referral Criteria

- **Diagnostic testing** (**Neurology referrals**)
  Late-onset progressive cerebellar ataxia and intention tremor
- **Family testing (Clinical Genetics referrals)**
  Relatives with a confirmed family history of an *FMR1* related disorder

Molecular Analysis

1) **FRAXA PCR Assay:** Fragment length analysis of *FMR1* gene [CGG]n triplet repeat. Pre-screen for the majority of diagnostic referrals.
2) **Asuragen Amplidex PCR Assay:** Fragment length analysis of *FMR1* gene [CGG]n triplet repeat expansions in pre-mutation and full mutation size range by CGG repeat primed PCR analysis.

Prices* & Turnaround Times (TAT)

* Prices available on request

Test	TAT
FMR1 expansion analysis (PCR)	42cd
FMR1 high sensitivity gene expansion analysis	42cd

Contact Details
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Accredited to ISO 15189:2012

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/
EDDNAL
http://www.eddnal.com/
OMIM
http://www.omim.org/
Genetic Test Registry
http://www.ncbi.nlm.nih.gov/gtr/
Support
www.fragilex.org.uk