Fragile X (A) syndrome (OMIM# 300624)

Background

Fragile X (A) is an X-linked disorder and the most common single-gene cause of inherited learning disabilities. Over 99% of cases are caused by an expansion of a CGG repeat in the 5'UTR of the gene *FMR1* (OMIM* 309550), resulting in methylation and silencing of the gene.

The condition predominantly affects males (approximately 1 in 4000-6000), where it causes moderate to severe intellectual and social impairment with syndromic features including large ears and head, long face and macroorchidism.

Females with a full expansion have a variable phenotype from phenotypically unaffected (~50% of cases) to moderate mental and social impairment.

Recommended Clinical Referral Criteria

- **Diagnostic testing (Paediatric or Clinical Genetics referrals)**
  - Diagnostic testing for patients with learning disability will usually only take place following a normal array CGH result. Please refer to the array CGH referral criteria for children with learning disabilities
- **Family testing (Clinical Genetics referrals)**
  - Relatives with a confirmed family history of Fragile X or FMR1 related disorder
- **Prenatal testing** is carried out at the Manchester Centre for Genomic Medicine (www.mangen.co.uk)

Molecular Analysis

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 high sensitivity gene expansion analysis	42cd

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Links

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

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.