

Fragile X syndrome **OMIM: 300624**
Gene: FMR1 **Locus: Xq27.3** **OMIM: 309550**

SERVICE: **PCR and Southern blot analysis for common expansion**

TESTING: **Diagnostic:** **clinically affected patients**
Carrier: **relatives of affected individuals (with known expansion)**
Prenatal: **at risk of having an affected child**

REFERRALS: **Clinical Geneticists and Paediatricians**
The laboratory does NOT accept referrals directly from patients

TARGET REPORTING TIME AND COSTS

(Non UK National Health Service patients are subject to a surcharge. Payment must be agreed prior to testing – please include invoice form A)

Diagnostic/	2 weeks	£145	(PCR only)
Carrier:	8 weeks	£175	(PCR and Southern blot)
Prenatal:	2 weeks	£350	(including maternal contamination studies)

TECHNICAL INFORMATION

- PCR and triplet-primed PCR analysis of the (CGG)_n triplet repeats within the 5' UTR (exon 1) of *FMR1*
- Southern blotting of the CGG repeat region of *FMR1* (methylation sensitive)

SAMPLE REQUIREMENTS

- 1-5ml blood in EDTA or 100ul DNA (concentration ~500ng/ul)
- All patient samples must be labelled with **name, date of birth and Hospital/NHS number**
- Samples should be accompanied by a FULLY completed request card (available from the laboratory)
- Please include details of test, family history, patient address & postcode, GP, referring clinician and unit/hospital
- **Samples and paperwork must include three unique and matching patient identifiers**

SHIPPING DETAILS

- DNA can be sent by first class post
- Blood must be appropriately packaged and preferably sent by courier to arrive as soon as possible.
- Do not freeze prior or during postage.

CONSENT

It is the responsibility of the referring clinician to ensure consent has been obtained for:

- testing and storage
- the use of the sample and the information generated from it to be shared with members of the patients family and their health professionals

After testing, part of this sample might be used anonymously for the development of new tests and to monitor the quality of laboratory results.

CONTACT DETAILS

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Reference No: 1275