UKGTN Testing criteria

UK Genetic Testing Network

Name of Disease(s): Fragile X Carrier, or premutation-test in relatives with known family history

Name of gene(s): Fragile X Mental Retardation 1 (FMR1)

Patient name: **Date of birth:**

Patient postcode: **NHS number:**

Name of referrer:

Title/Position:
Lab ID:

Referrals will only be accepted from one of the following:

Referrer	Tick if this refers to you.
Clinical Geneticist / Genetic counsellor	
Obstetrician / fetal medicine specialist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:

Criteria	Tick if this patient meets criteria
Known family history of FraX, premutation FraX, or intermediate allele of uncertain stability and ≥ 56 repeats (but excluding generationally stable intermediate allele), **AND** potentially sharing same X chromosome.	
OR Female with close family history of male relative with severe learning difficulty without specific diagnosis, especially if he fits guidance notes for affected male (see specific dossier).	

If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.

Approval Date: December 2008