UKGTN Testing criteria

UK Genetic Testing Network

Name of Disease(s): Fragile X Mental Retardation Syndrome (Full Mutation in Males)

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.
Paediatrician, Community Paediatrician or Paediatric Neurologist	
Child Psychiatrist	
Clinical Geneticist / Genetic Counsellor	
Learning disability consultant / Psychiatrist/ Neurologist	

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

Criteria	Tick if this patient meets criteria
Male	
AND Moderate to severe Developmental delay / learning difficulty (IQ if measured would be 35-70 range)	
AND Does not have profound psychomotor handicap necessitating total care	
AND FraX test specifically requested by referrer (ie. not laboratory-initiated)	

Guidance notes:

Typically will have:
Gaze Avoidance
Resists physical contact (tactile defensiveness)
Repetitive speech/ perseveration (delayed echolalia) & behaviours on autistic spectrum (hand-flapping, hand biting)
Inattentive & distractable; motor hyperactivity/ restlessness
Shy & socially anxious, but also sociably friendly
Particular difficulty with numeracy and visuo-spatial skills
Head circumference > 50th centile
Joint laxity

Continued on next page……

Guidance notes cont:

Also may have:
Family history of learning difficulty
Long ears-post- 7 yrs
Large testes-post-pubertally
Epilepsy

Very unlikely to have:
High functioning (normal intelligence) autism/ Asperger syndrome
Classic 'aloof' severe autistic phenotype
Microcephaly

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.