UKGTN Testing criteria

UK Genetic Testing Network

Name of Disease(s): Fragile X Mental Retardation Syndrome (Females as index case)

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
Female	
AND learning difficulty (usually mild, IQ often 80-85; but can be moderate or severe LD)	
AND Does not have profound psychomotor handicap necessitating total care	
AND FraX test specifically requested by referrer (ie. not laboratory-initiated)	

Guidance notes:

Often will have:
Attention difficulty
Gaze avoidance
Resists physical contact (tactile defensiveness)
Communication problems, impulsive social faux pas in speech
Shy, self-conscious & socially anxious behaviour
Obsessionality
Passivity
Particular difficulty with maths and visuo-spatial skills
Poor adaptive skills / vulnerable

Continued on next page……

Guidance notes cont:

Also may have: Family history of learning difficulty
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.