UKGTN Testing criteria

UK Genetic Testing Network

Name of Disease(s): LUBS X-LINKED MENTAL RETARDATION SYNDROME; MRXSL (300260)
Name of gene(s): Contiguous gene duplication / triplication of Xq28 (including MECP2 and IRAK1) methyl CpG binding protein 2 (Rett syndrome); MECP2 (300005) interleukin-1 receptor-associated kinase 1; IRAK1 (300283)

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.
Consultant clinical geneticists	

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

Criteria	Tick if this patient meets criteria
ESSENTIAL CRITERIA	
Male	
Sporadic case or X-linked inheritance	
Severe mental retardation	
Initial hypotonia	
AND in infancy evidence of X-linked inheritance	
AND at least one MINOR CONGENITAL CRITERIA (apparent at birth)	
Dysmorphic appearance	
Microcephaly	
Undescended testicles	
And at least 2 of LATER MAJOR CRITERIA (These criteria are unlikely to be present at birth)	
Progressive spasticity	
Absent speech	
Recurrent infection	
OR relative at-risk in family with proven mutation	

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.