Why MedGenome

- Unmatched experience of 350,000+ genomes/exomes
- South Asia largest database with 5,000,000+ variants
- Recommended by 4000+ hospitals & trusted by 10000+ clinicians
- Consistent proficiency testing through "External Quality Monitoring Programs"
- Two levels of analysis and review by molecular and clinical geneticists

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MEDGENOME | ACTIA Inherited Disease Genetics

Fragile X Syndrome

Fragile X Syndrome (FXS)

- Most common genetic cause of inherited intellectual disability and autism spectrum disorder (ASD).
- Early identification results in appropriate management and improvement in functioning. Risk assessment in other family members can lead to prevention of the disorder.
- Estimated incidence is 1 in 4000 males and 1 in 8000 females (Hagerman RJ., 2017)
- 99% of FXS is primarily caused due expansion of a segment of CGG repeats in the 5' UTR of FMR1

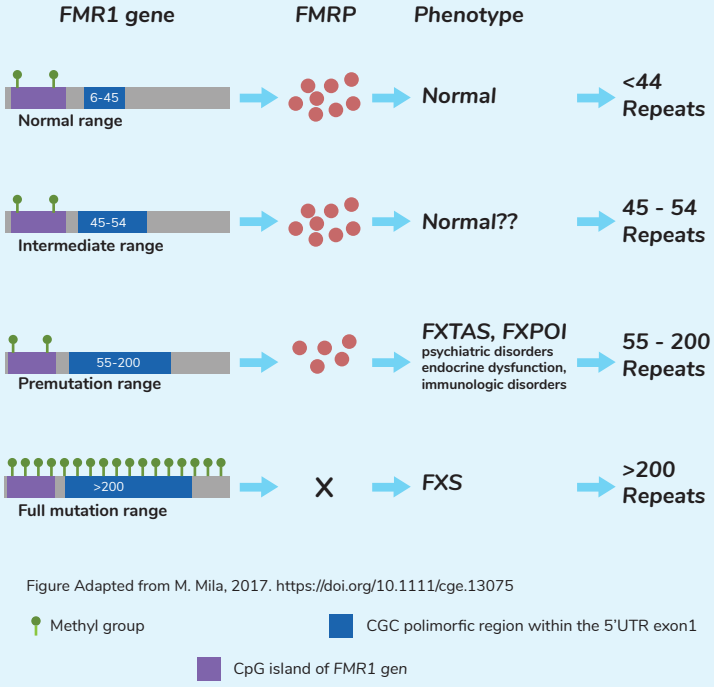


Figure Adapted from M. Mila, 2017. https://doi.org/10.1111/cge.13075

Methyl group

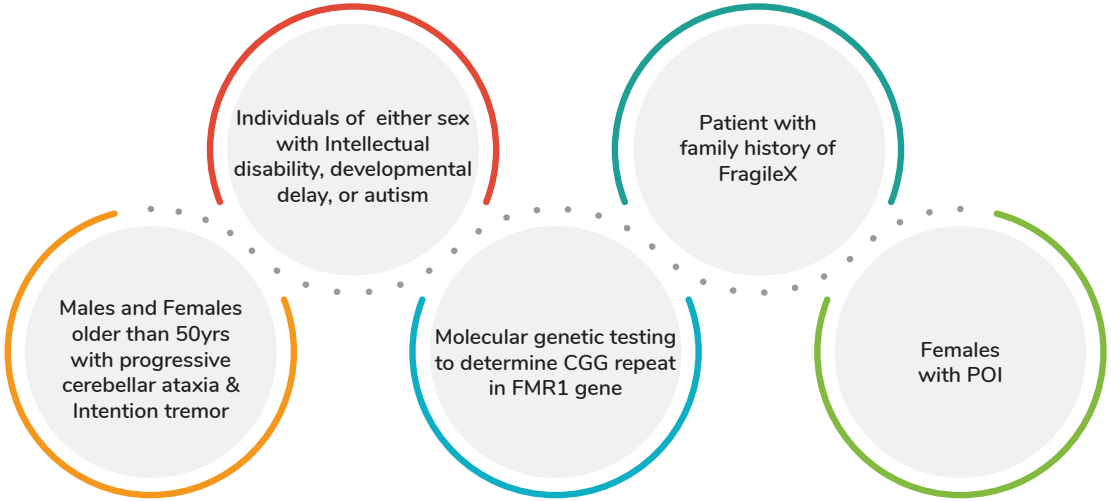
CGC polimorfic region within the 5'UTR exon1

CpG island of FMR1 gen

Key Features of this PCR(TP-PCR) based Assay:

- Accurately sizes alleles upto 200 CGG repeats
- Resolves and confirms heterozygous alleles in Females
- Validated in positive cases and controls with known repeat expansion
- Repeatability, reproducibility, sensitivity and specificity tested

Who Should Be Tested for Fragile X Syndrome:



Several medical professional societies including American Academy of Pediatrics Committee on Genetics have recommended offering chromosomal microarray (CMA) testing and Fragile X testing for individuals with ASD

Consensus Statement of the Indian Academy of Pediatrics on Diagnosis and Management of Fragile X Syndrome in India (2019)

Test Details

Test Code	Test Name	Sample Type	TAT
MGM1221	Fragile X Syndrome by PCR for Male	Peripheral blood in EDTA	21 Working Days
MGM1222	Fragile X Syndrome by PCR for Female	Peripheral blood in EDTA	21 Working Days

Reference PMID: 28960184, 33795824, 25227148