ACTIA
Inherited Genetics

MEDGENOME

Fragile

Syndrome

- Fragile X syndrome (FXS) is a genetic disease caused by alterations in *FMR1* (fragile X mental retardation 1) gene sequence.
- Estimated incidence is 1 in 4000 males and 1 in 8000 females (Hagerman RJ., 2017)
- Around 30% of males with FXS meet criteria for autism. Approximately 50% of female carriers will have mild to moderate mental disabilities.
- 99% of FXS is primarily caused due expansion of a segment of CGG repeats in the 5' UTR of FMR1

Fragile X Syndrome (FXS) is the 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. This necessitated the formulation of IAP recommendations for the diagnosis and management of FXS in Indian children and adolescents.

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

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

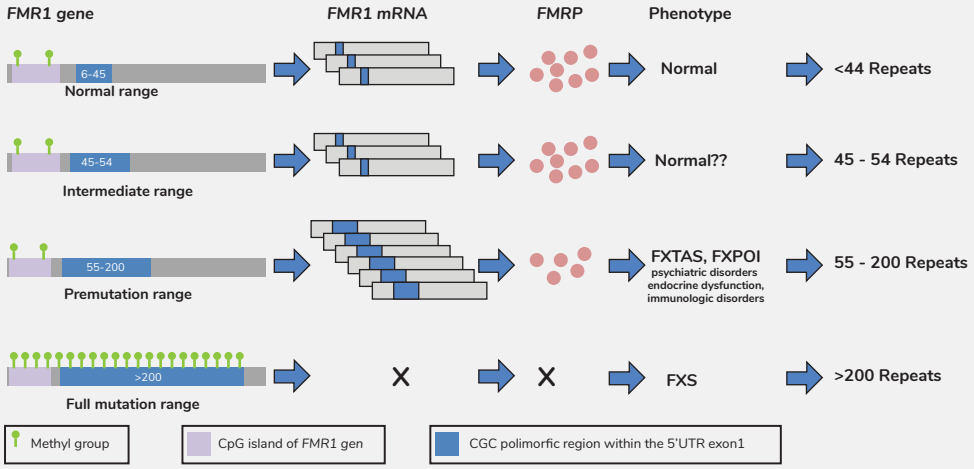
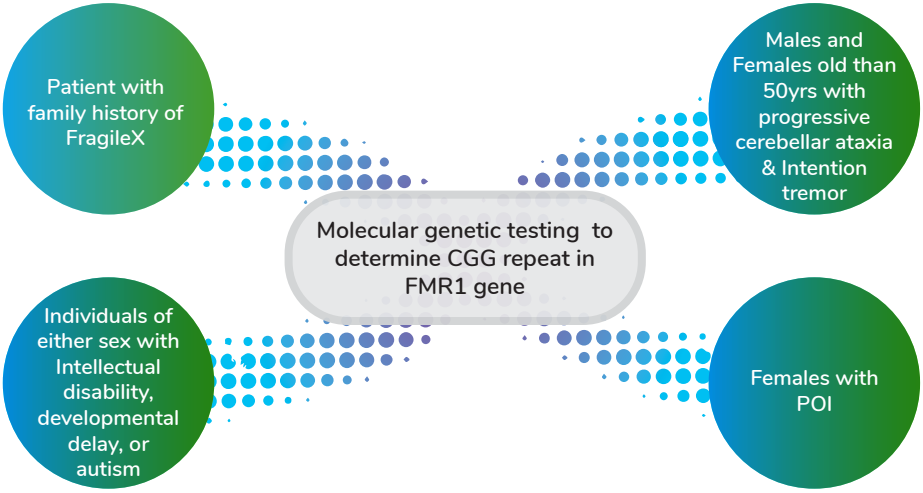


Figure Adapted from M. Mila, 2017. https://doi.org/10.1111/cge.13075

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

Test Code	Test Name	Sample type recommended	
MGM1221	Fragile X Syndrome by PCR for Male	Peripheral blood in EDTA	
MGM1222	Fragile X Syndrome by PCR for Female	Peripheral blood in EDTA	

Reference PMID: 28960184, 33795824, 25227148

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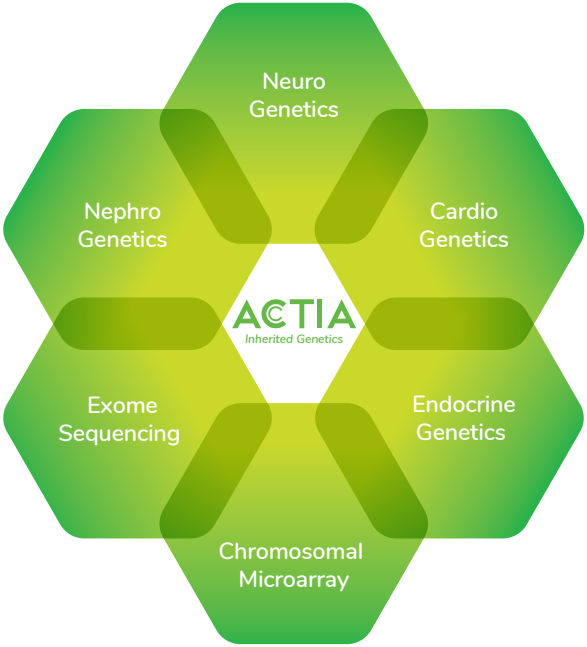
Actia from MedGenome provides end-to-end integrated solution for clinical genomics in India



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