LABORATORY REPORT

Redcliffe labs

Patient NAME	:		
DOB/Age/Gender	:	Report STATUS	:
Patient ID / UHID	:	Barcode NO	:
Referred BY	:	Sample Type	: Whole blood EDTA
Sample Collected	: Aug 25, 2025, 01:58 PM.	Report Date	: Sep 08, 2025, 01:38 PM.

#Fragile X (FMR 1) Mutation Screen

CLINICAL DETAILS: ASD/ HA, delayed speech, poor understanding, poor socialization. ?Fragile X syndrome.

TEST RESULTS :

Result Summary	Normal
Number of CGG repeats	38

Interpretation :

Analysis of the pathogenic (CGG)n trinucleotide repeat region of FMR1 gene revealed presence of alleles in the normal range.

Test Information :

Methodology: PCR followed by agarose gel electrophoresis.

Fragile X syndrome is caused by an expansion of CGG repeat sequences in the FMR1 gene in 99% of the cases. The interpretation is based on the following types of repeat sequences:

1. Normal < 45 repeats
2. Intermediate 45-54 repeats
3. Pre-mutations 55-200 repeats
4. Full mutation > 200 repeats

Comments: A further correlation with clinical findings and history is recommended. Conformation with fragment analysis based assay is suggested, if clinically indicated.

Notes:

Rare FMR1 mutations unrelated to trinucleotide expansion, intellectual disability associated with other fragile X sites, in particular FRAXE, or other gene mutations, methylation studies of FMR1 gene are not detected by this assay. Possible diagnostic errors include sample mixups and genotyping errors resulting from trace contamination of PCRs, maternal cell contamination of fetal samples and from rare polymorphisms, which interfere with analysis. Low level mosaicism may not be detectable by this method. Mutation analysis should be combined with phenotypic, cytogenetic, microarray analysis and pedigree data for the most accurate interpretation.

Acceptable limitations of size analysis :

consensus size ± 5 repeats for alleles with <55 repeats; consensus size ± 10 repeats for alleles with 56-100 repeats; and consensus size ± 2 SDs for alleles with >100 repeats.

Approved by
Dr. Himani Pandey
Postdoc-SGPGIMS Lucknow
Lab Head-Clinical Genomics

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Reference:

Saluto, Alessandro, et al. "An enhanced polymerase chain reaction assay to detect pre-and full mutation alleles of the fragile X mental retardation 1 gene." The Journal of Molecular Diagnostics 7.5 (2005): 605-612.

NOTE- **This test is processed at Redcliffe's partnered lab.

*** End Of Report ***

Approved by
Dr. Himani Pandey
Postdoc-SGPGIMS Lucknow
Lab Head-Clinical Genomics

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