PROGENE LAB
For Medical And Genetic Tests
معمل بروجين للتحاليل الطبية

Genetic Analysis Report for Fragile X Syndrome

Patient Name: **********	**اسم المريض:** **********
Sample type: Blood sample	**DOB:** **********
Phone No.: **********	**Sex:** Male
Referring Doctor: **********	**City, Country:** Egypt
Report type: Analysis for Fragile X Syndrome	**Order received:** **********
Report date: **********	
Reason of referral: Suspicion of genetic etiology based on clinical features	

Clinical Information: A 16-year-old male child complaining of intellectual disability and speech delay with suspicion of fragile X syndrome.

Summary of The Results

Positive result

An expanded number of the triplet CGG repeats copies which falls in the region of a premutation

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تليفون: 01101799014 – 01149552098

Interpretation of the test results:

Gene	Phenotype	Heredity	Allele 1	Normal	Intermediate	Premutation	Pathogenic
FMR1 (OMIM* 30955)	FXS (OMIM# 300624)	XLD	**155**	5-44	45-54	55-200	>200 repeats

The patient will have clinical features and developmental delay of Fragile – X Syndrome but in a milder form than the full mutation.

Recommendation:

- Genetic counseling should be offered with all diagnostic genetic testing.

Methods:

Methods Repeat analysis: A repeat spanning polymerase chain reaction (PCR) was performed to determine the number of CGG repeats in the 5'-UTR region of the gene FMR1. Additionally, a repeat-primed PCR method specifically designed to detect larger repeat expansions was performed. This was followed by separation and sizing of the PCR fragments by capillary electrophoresis. The test was performed according to the standard protocol of the AmplideX®-FMR1-mPCR method.

Please be aware that due to the possibility of rare variants in a primer binding site, the unlikely event of an allelic dropout cannot be excluded for molecular diagnostics based on PCR technology. The sample fulfilled our quality criteria upon arrival and during/after each processing step in the laboratory.

With kind regards

Clinical Laboratory Geneticists Staff:

Prof. Dr. Marwa Elsharkawy

Associate Prof. Noha Abd Elhalim

Signature

Prof. Dr. Mohamed Abd Elmonem

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