Asuragen Clinical Services
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Patient and Order Information

<Date>

Patient Name:	DOB:	Sex: <M F>
Order ID:	Sample ID: AG651	Type of sample: <gDNA/whole blood>
MRN:	Date collected:	Date received:
Attending Provider:	Clinical History:	

Results

Allele	*5'UTR CGG Repeats*	*Genotype*	*Consecutive CGG Repeats**
Allele1	**35**	**(CGG) 35**	**35**
Allele2	**45**	**(CGG) 9 (AGG) (CGG) 9 (AGG) (CGG) 25**	**25**

*Consecutive CGG repeats are provided as the largest value detected for each *FMR1* allele.

Additional Comments

Background and Significance

Expansions of the CGG triplet repeat in the 5'-untranslated region of the fragile X mental retardation-1 (*FMR1*) gene are associated with several disorders, including fragile X syndrome, fragile X-associated tremor/ataxia, and fragile X-associated primary ovarian insufficiency. Many *FMR1* alleles contain AGG sequences that are interspersed among the CGG repeats, usually in the 5' portion of the repeat region. Published literature suggests that these AGG interruptions confer DNA stability, and may reduce the risk of expansion in the next generation (Eichler 1994; Kunst 1994; Zhong 1996; Nolin 1996). Premutation alleles with as few as 56 and 59 CGGs and lacking AGG interruptions have been shown to expand to full mutations (>200 CGGs) within a single generation (Nolin 2003; Fernandez-Carvajal 2009).

Assay description and methodology

This laboratory developed test consists of PCR amplification of the 5'-untranslated region of the *FMR1* gene followed by capillary electrophoresis size analysis of the amplified products. Determination of the comprehensive *FMR1* genotype is accomplished from fragment analyses of allele-specific amplicons that report both the number and sequence context of AGG sequences that interrupt the CGG repeat tract.

Jennifer Skeen, Ph.D, CG(ASCP)
Director, Pharmacogenomic and CLIA Laboratory Operations

Date

Anna (Szafranska) Schwarzbach, MSc, PhD, MB(ASCP)^{CM}, CQ
CLIA Laboratory Manager

Asuragen recommends that a trained genetics professional discuss these results with the patient. If no genetic services are available, please contact Asuragen for access to a board-certified Genetic Counselor.

Disclaimer

This test was developed and its performance characteristics determined by Asuragen's Clinical Laboratory. It has not been cleared or approved by the US Food and Drug Administration. FDA does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical laboratory testing. This test cannot differentiate samples containing a single allele from samples containing multiple alleles with identical genotypes.

Confidentiality of this medical record shall be maintained except when use or disclosure is required or permitted by law, regulation, or written authorization by the patient.

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