Xpansion Interpreter®

Delivering clear genotype and AGG interruption information for the *FMR1* gene to help clarify risk of expansion to full mutation

Xpansion Interpreter® is the first clinically available laboratory-developed *FMR1* test to determine the total number of CGG repeats and number of AGG interruptions. The number of AGG interruptions provide insight into how likely select premutation carriers are of having a child with fragile X syndrome (FXS). FXS affects approximately 1 in 3,600 – 4,000 males and 1 in 4,000 – 6,000 females[1] worldwide and is the most common inherited cause of intellectual disability and autism.

WHO TO TEST

Use the criteria below to identify patients who may be appropriate for testing with Xpansion Interpreter:

- Known intermediate and premutation fragile X carriers (50-90 CGG repeats)
- Family history of fragile X or fragile X-related disorders[2]
- Family history of unexplained intellectual disability, developmental delay or autism
- Personal or family history of female infertility
- Relevant fragile X-related disorders including fragile X-associated primary ovarian insufficiency (FXPOI) and fragile X-associated tremor/ataxia syndrome (FXTAS)

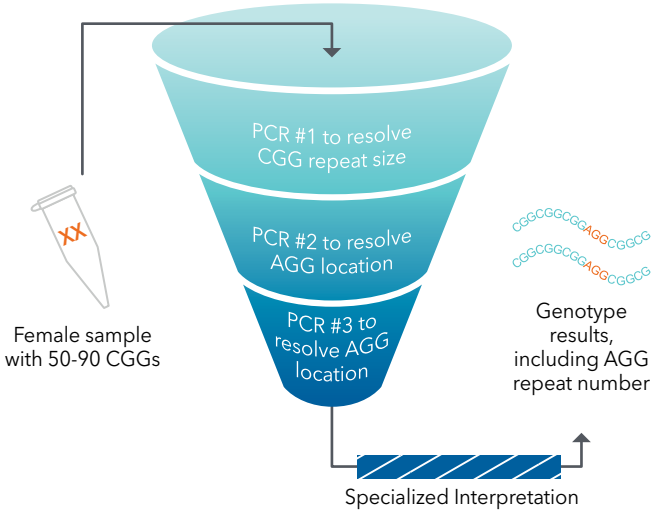
CATEGORIES OF FRAGILE X MUTATIONS

CATEGORY	NUMBER OF REPEATS	CLINICAL FEATURES
Normal	<45	• Normal • Offspring have normal-size gene region
Intermediate/ Gray zone (unaffected)	45-54	• Normal • Not at risk for passing a full mutation to next generation • **Possible candidate for Xpansion Interpreter (50-54 repeat range)**
Premutation (carrier)	55-200	• Typically intellectually normal • Females at risk for having children with fragile X syndrome • **Strong candidate for Xpansion Interpreter (55-90 repeat range)**
Full mutation	>200	• >99% males and most females have clinical features of fragile X syndrome

Asuragen®
a biotechne® brand

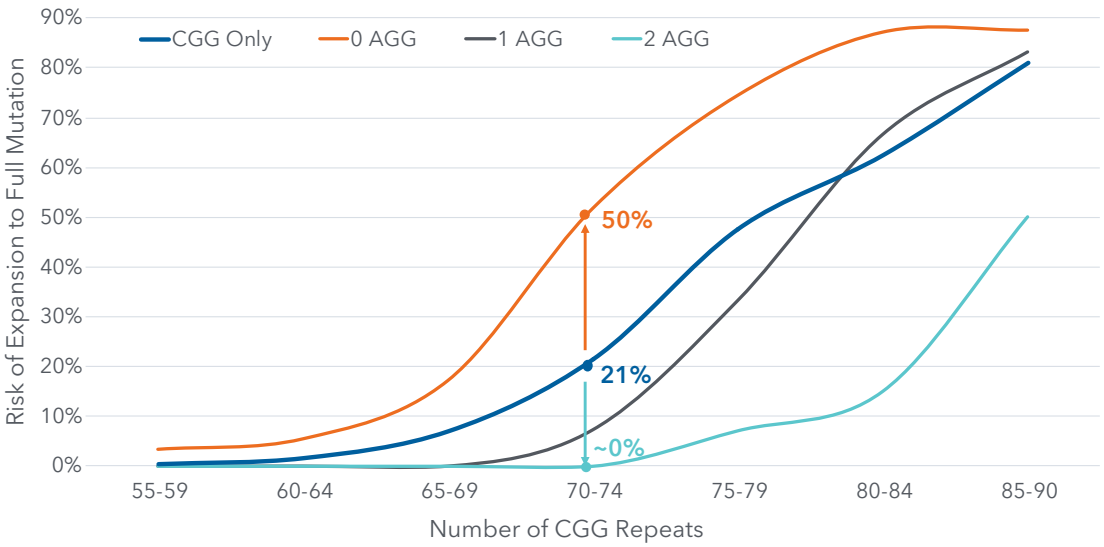
PROPRIETARY DESIGN ENSURES HIGH-QUALITY GENOTYPE RESULTS

Asuragen's Xpansion Interpreter® testing service employs three different PCR reactions for each DNA sample. Through specialized training to interpret the results from all three PCR reactions, the Asuragen CLIA laboratory is able to definitively determine the location of the AGG interruptions within CGG repetitive sequences. Xpansion Interpreter results include reporting of allele-specific percent risk of expansion based on the detected genotype.



AGG INTERRUPTION QUANTIFICATION STRATIFIES RISK[3]

AGG interruptions provide important information beyond that provided by CGG repeats alone (blue line). For example, for a woman with a 70 CGG repeat allele, risk of expansion to a full mutation can range from ~0% to 50% depending on the number of AGG interruptions. **To provide the best information for reproductive decisions, determining number of AGG interruptions is vital**.



CONTACT US

Xpansion Interpreter (AGG interruption testing) is provided as a service to specific carrier screening laboratories throughout the United States. If you would like information on how to obtain XI, please contact us: 1.877.772.8018 or aus.clinicallabsupport@bio-techne.com.

Do not send CONFIDENTIAL Patient and Protected Health Information to this email address. Please send all Patient and Protected Health Information to Asuragen via fax at 1.512.681.5205 or call Asuragen Client Services at 1.877.772.8018.

For more information and published articles, please visit: asuragen.com/portfolio/genetics/xpansion-interpreter/

Xpansion Interpreter® is a Laboratory Developed Test (LDT). The analytical and clinical performance characteristics have not been reviewed or approved by the U.S. FDA.

References: 1. National Fragile X Foundation. 2021. ; 2. Monaghan KG, et al. *Genet Med*. 2013.; 3. Nolin SL, et al. *Genet Med*. 2015.; Nolan SL, et al. *Am J Med Genet A*. 2013.

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Asuragen, Inc.
2150 Woodward Street, Suite 100
Austin TX 78744
www.asuragen.com

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