GENETICS & WOMEN'S HEALTH

One Fast Result for Fragile X Risk Assessment

What do we mean by one fast result? One lab, one report, one simultaneous and precise result for fragile X syndrome.

AGG analysis in women who have a premutation with 55-90 CGG repeats provides a more accurate risk assessment compared to CGG testing alone.[1-3] **And with our unique, integrated approach, this combined CGG/AGG fragile X result is available without extending turnaround time, providing you and your patients with one timely and more refined risk estimate.**

- More precise fragile X risk information – enables more informed reproductive planning and enhanced medical decision-making for at-risk pregnancies
- Significant turnaround time savings – more convenient for you and less anxiety for your patients due to simultaneous CGG and AGG reporting from our lab
- Available for Inheritest Carrier Screen panels and our single-gene fragile X tests

Inheritest® Carrier Screen
Lab report including a final CGG/AGG fragile X result

Expected turnaround time: 14-21 days

Inheritest Carrier Screen offers a fast turnaround time for a complete and final fragile X result with both CGG and AGG repeats reported.

labcorp

Fragile X carriers and risk of expansion

Fragile X syndrome occurs in individuals with greater than 200 CGG repeats. The risk of expansion in premutation carriers (55-200 CGG repeats) varies depending on the number of repeats.[4]

In premutation carriers with repeat numbers between 55-90 CGGs, the number of AGG interruptions influences risk of expansion to a full mutation. The presence of AGG interruptions is associated with a lower risk of expansion to a full mutation.[4]

Risk of expansion to a full mutation based on CGG repeat size and AGG data[4]

Maternal CGG repeat size range*	0 AGGs	1 AGG	2 or more AGGs	
55-59	1.90%	<1%	<1%	**Example:** In a patient with 75-79 CGG repeats, the risk of expansion to a full mutation is 10.7% for 2 AGG interruptions compared to 71.7% for no AGG interruptions.
60-64	5.40%	<1%	<1%	
65-69	10%	<1%	<1%	
70-74	51.90%	7.60%	<1%	
75-79	**71.70%**	**40%**	**10.70%**	
80-84	88.20%	65.20%	20.70%	
85-90	86.10%	84.60%	29.40%	

*AGG analysis is not performed for CGG repeats >90 because once the repeat length exceeds this number, there is no apparent effect of AGG interruptions.[3]

Fragile X syndrome test offering

Test/Panel Name	Test No.
Inheritest® Core Panel	**481776**
Inheritest® 14-gene Panel	**481797**
Inheritest® High Frequency Panel	**481816**
Inheritest® 100 PLUS Panel	**481855**
Inheritest® 300 PLUS Panel	**481874**
Inheritest® 500 PLUS Panel	**481893**
Fragile X Syndrome, Carrier	**481684**
Fragile X Syndrome, Diagnostic	**481701**

Specimen requirements:
Blood: 8.5 mL whole blood in a yellow-top (ACD-A) tube (preferred) or lavender-top (EDTA) tube; yellow-top (ACD-B) tube is not acceptable.

Saliva: Saliva specimens are accepted in Oragene® Dx saliva collection kits. To order saliva kits, please contact your sales representative or Client Services at 800-848-4436.

Buccal swab: Specimens collected using buccal swab kits are accepted for Fragile X tests and Inheritest Core Panel. To order buccal swab kits, please contact your sales representative or Client Services at 800-848-4436.

References
1. Yrigollen CM, Durbin-Johnson B, Gane L, et al. AGG interruptions within the maternal FMR1 gene reduce the risk of offspring with fragile X syndrome. *Genet Med*. 2012. 14(8):729–736.
2. Nolin SL, Sah S, Glicksman A, et. Al. Fragile X AGG analysis provides new risk predictions for 45-69 repeat alleles. *Am J Med Genet* Part A 2013. 161A:771-778
3. Nolin SL, Glicksman A, Ersalesi N, et al. Fragile X full mutation expansions are inhibited by one or more AGG interruptions in premutation carriers. *Gen Med*, 2015 May;17(5):358-64.
4. Domniz N, Ries-Levavi L, Cohen Y, et al. Absence of AGG Interruptions Is a Risk Factor for Full Mutation Expansion Among Israeli FMR1 Premutation Carriers. *Front Genet*. 2018. 9:606.

Call Us
Toll-free (within the U.S.): 800-848-4436

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WHGeneticsCS@Labcorp.com

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Labcorp
3400 Computer Drive
Westborough, Massachusetts 01581

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