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

Inheritest® Carrier Screen **Lab report including a final CGG/AGG fragile X result**	~ 14 days

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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	1.9%	<1%	<1%
60-64	5.4%	<1%	<1%
65-69	10%	<1%	<1%
70-74	51.9%	7.6%	<1%
75-79	**71.7%**	**40%**	**10.7%**
80-84	88.2%	65.2%	20.7%
85-90	86.1%	84.6%	29.4%

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.

*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 number	Test/panel name	Turnaround time	Specimen requirements
451964	Inheritest Carrier Screen, *Core* Panel	9-14 days	10 mL whole blood for all testing options
451960	Inheritest Carrier Screen, *Society-guided* Panel	11-14 days	
451920	Inheritest Carrier Screen, *Ashkenazi* Jewish Panel	11-14 days	
451950	Inheritest Carrier Screen, *Comprehensive* Panel	12-16 days	
523/511919 (IG/LabCorp)	Fragile X, PCR With Reflex to Southern Blot	6-14 days	
521/511655 (IG/LabCorp)	Fragile X, PCR and Southern Blot Analysis	8-14 days	

REFERENCES

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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.

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