AmplideX®

Fragile X Dx & Carrier Screen Kit

Fragile X syndrome (FXS) is the most common inherited form of intellectual disability and autism, affecting approximately 1 in 4,000 males and 1 in 8,000 females in the United States.[1] The disorder is X-linked and is caused by a full mutation expansion (>200 CGG repeats) within the *FMR1* gene. Testing for fragile X syndrome and its associated disorders (FXTAS, FXPOI) necessitates the accurate sizing of *FMR1* CGG repeats across distinct clinical categories: normal (5-44 repeats), intermediate (45-54 repeats), and premutation (55-200 repeats).

The AmplideX® Fragile X Dx & Carrier Screen Kit is an in vitro diagnostic device intended as an aid in the post-natal diagnosis of fragile X syndrome, and fragile X-associated disorders and for carrier testing in adults of reproductive age. Based on Asuragen's unique, PCR-only approach, the assay reliably amplifies and detects all alleles, including low-level mosaics and full mutations, in a simple, streamlined workflow, which includes automated software. Requiring only 20ng of DNA from whole blood, the assay provides results within one day.

REDUCED COMPLEXITY

Ease of data analysis and reporting

- Cleared test supports rapid assay validation
- Implementation of proprietary PCR solution for amplifying GC-rich regions
- Clinically-validated AmplideX PCR/CE Fragile X Reporter software automates sample genotyping

OPTIMIZED WORKFLOW

Reduces valuable operator hands-on-time and overall turnaround time

- A single multi-allele control provides a peak in every clinical category and can be used as positive control
- Up to 50-fold reduction in Southern blot analysis
- End-to-end solution for *FMR1* analysis including all necessary reagents and software

QUALITY RESULTS

Highly-sensitive, precise, and accurate assessment of allele size for screening and diagnosis

- Detection of challenging allele expansions — including low abundance full mutation size mosaics — provides more sensitive and accurate diagnosis of Fragile X
- Rapid and accurate sizing enables high throughput identification of premutation carriers; access to Xpansion Interpreter® can further refine the risk to full mutation expansion
- Proven performance of technology as indicated by over 100 peer-reviewed publications

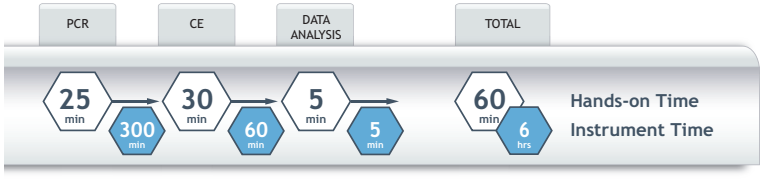

Figure 1. Workflow for the AmplideX® Fragile X Dx & Carrier Screen Kit

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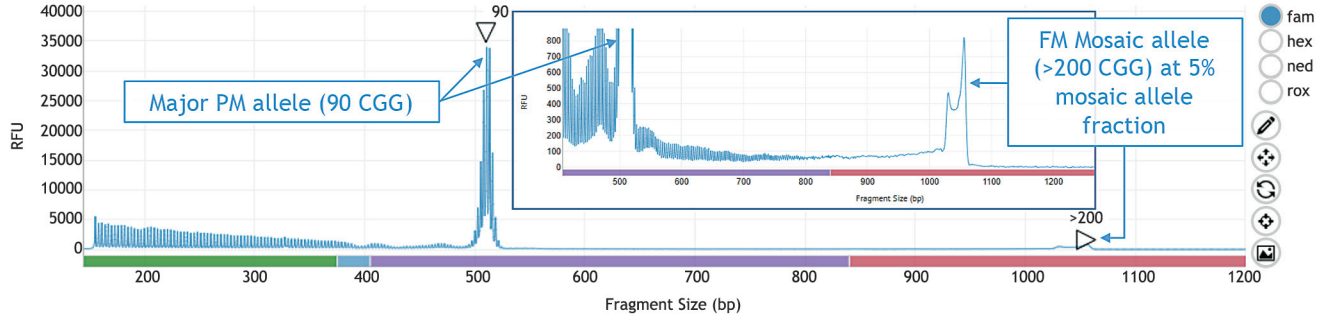

Figure 2. Example of major premutation (PM) allele (90 CGG) with full mutation (FM) mosaic allele (>200 CGG) at 5% mosaic allele fraction

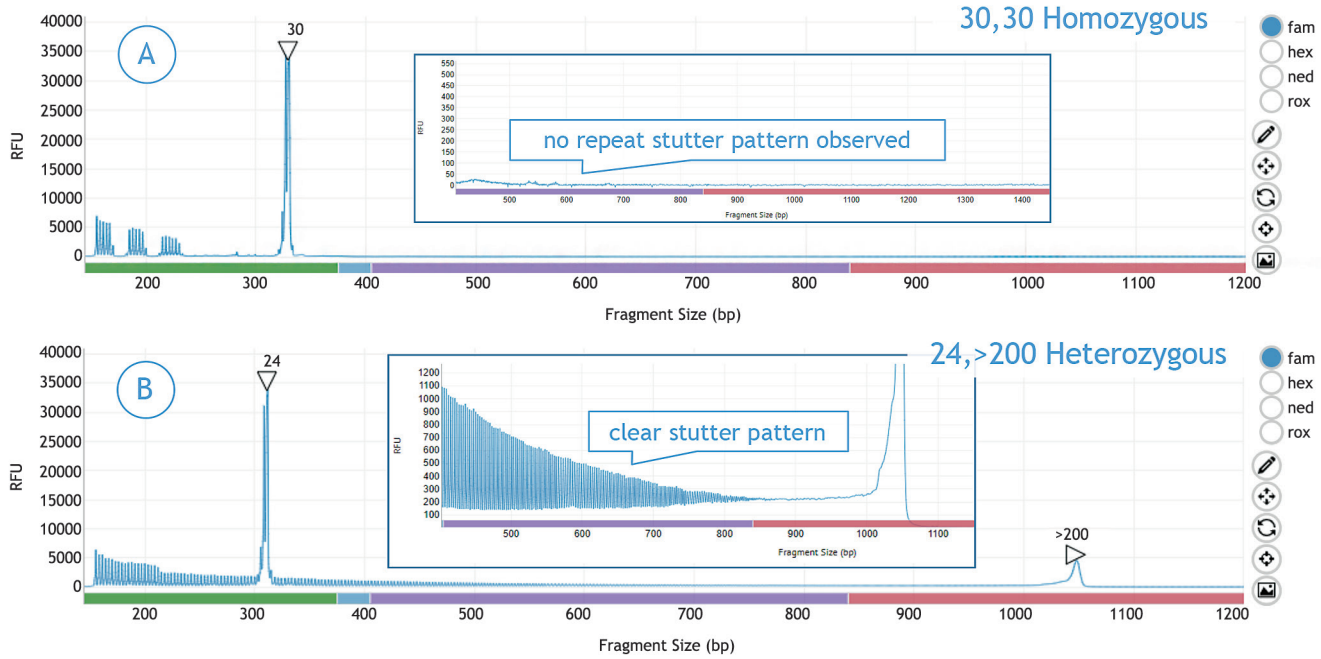

Figure 3. Clear, visual resolution of zygosity via repeat primer ("stutter") peak pattern. A) Shows a sample with a homozygous 30/30 CGG call. No stutter pattern is present after the gene-specific peak, indicating no further peaks are present. B) Shows a heterozygous sample with a heterozygous 24/> 200 CGG call. There is a clear stutter peak pattern after the first gene-specific peak (24), indicating the presence of another gene-specific peak (>200).

Product Name	Number of Reactions	Catalog Number
AmplideX Fragile X Dx & Carrier Screen Kit	100	49591

Asuragen®

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1. National Organization for Rare Diseases (NORD)