HUMAN HEALTH

ENVIRONMENTAL HEALTH

FRAGILE X MOLECULAR DIAGNOSTICS

FragilEase™

Brochure not for distribution in the USA.

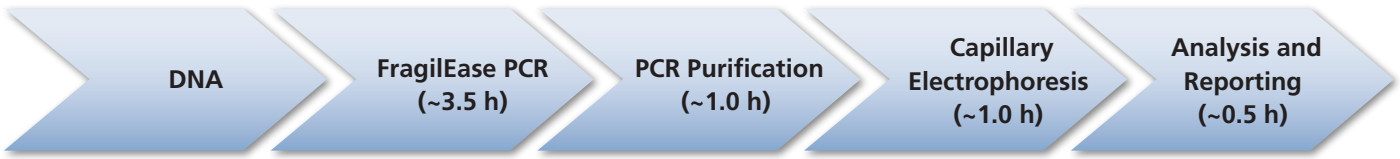
PerkinElmer®
For the Better

FragilEase™

Fragile X Syndrome is one of the most commonly inherited forms of intellectual disability and is caused by trinucleotide repeat expansions in the promoter region of the *FMR1* gene on chromosome X. When the CGG motif expands above 200 repeats, the *FMR1* gene is silenced by methylation causing Fragile X Syndrome with its typical characteristics including elongated face, large or protruding ears and intellectual disability.

FMR1 alleles are typically categorized into normal, premutation and full mutation alleles based on the number of CGG repeats. While Fragile X syndrome is the most severe form of the disease, individuals with repeat expansions in the premutation range of 55 – 200 repeats can be affected by Fragile X-associated tremor/ataxia syndrome (FXTAS), or Fragile X-associated primary ovarian insufficiency (FXPOI).

Traditionally, Fragile X testing is performed by use of a laboratory developed *FMR1*-specific PCR often followed by capillary electrophoresis. However, amplifying the entire CGG-rich template beyond about 100 – 130 repeats is challenging. In addition, differentiating full mutations from homozygous normal female samples has historically required a Southern blot reflex test.



PerkinElmer's FragilEase™ PCR assay is designed to amplify the entire CGG repeat sequence in the *FMR1* promoter region. By use of proprietary PCR reagents that allow an accurate amplification of the trinucleotide repeats, FragilEase™ can reliably detect full mutations with over 900 repeats hence significantly reducing the amount of Southern blot reflexing. In addition, together with the LabChip MultiDX electrophoresis instrument and FraXsoft™ for data analysis and interpretation, PerkinElmer offers a streamlined workflow solution for quick and easy sample analysis with a reporting time of less than a day.

LabChip MultiDX

By using PerkinElmer's patented microfluidic technology to perform reproducible, high-resolution electrophoretic separations the LabChip MultiDX provides a rapid, accurate and cost-effective way of analyzing FragilEase™ derived PCR products in a high throughput manner. The LabChip MultiDX microfluidics platform is supported by the 5K Chip and 5K Reagents Kit that are designed to provide the user with everything needed to analyze the FragilEase™ PCR products.

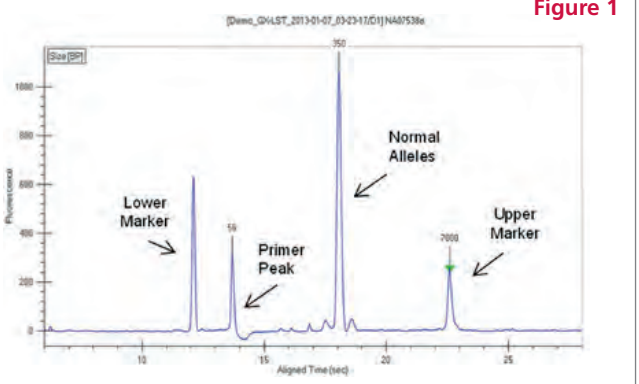
LABCHIP MULTIDX SPECIFICATIONS

- Height 18.75 in. (48 cm)
  Width 19.25 in. (49 cm)
  Depth 25.375 in. (64 cm)
  Weight 88 lbs. (40 kg)
- Plate formats: 96-well or 384-well microplates
- Weight 40 kg (88 lbs)
- Operating Temperature 65° to 78°F (18° to 26°C)
- Operating Humidity 20% to 70% relative humidity, noncondensing
- Power Input: 3.0A max at 100-127Vac, 50-60Hz (±10%); 1.5A max at 200-240Vac, 50-60Hz (±10%)

Features

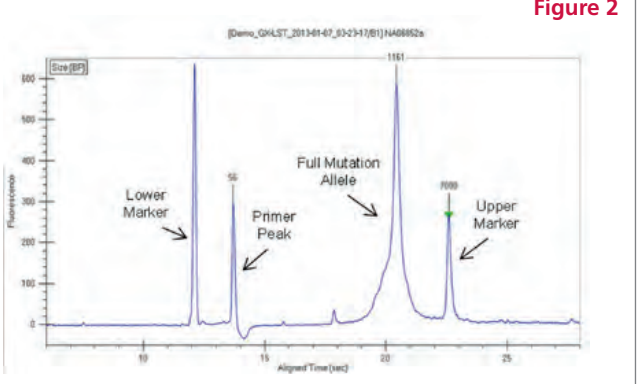
- Compact Benchtop Footprint
- High throughput analysis using 96- and 384-well plates
- CE marked for IVD use

Figure 1



Microfluidic capillary electrophoresis analysis of a Coriell female normal sample (NA07538).

Figure 2



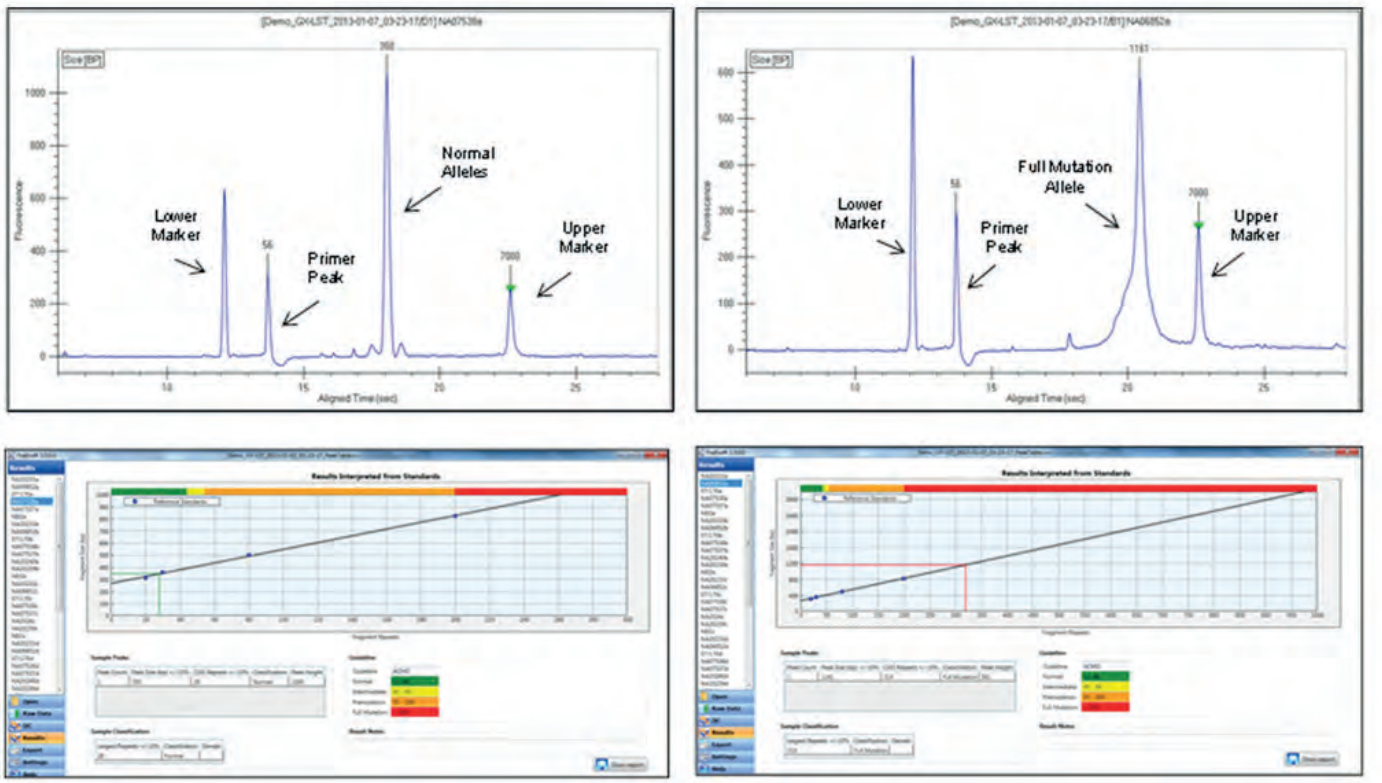
Microfluidic capillary electrophoresis of a Coriell male Fragile X (NA06852) with >200 repeats.

FraXsoft™

FraXsoft™ provides an easy-to-use analysis and interpretation option for our FragilEase™ products. FraXsoft™ allows the analysis of data derived from various microfluidic capillary electrophoresis systems and, based on the number of repeats in user-defined standard samples, calculates the number of CGG trinucleotide repeats in the samples tested. The mutation status of each sample can be classified according to ESHG, ACMG, or user-defined guidelines. FraXsoft™ provides intuitive data visualization for streamlined, quick and clear data interpretation of FragilEase™ data.

FRAXSOFT™ CHARACTERISTICS

- Repeat size calculation based on user-defined standard samples
- Graphical as well as numerical display of data based on ESHG, ACMG or user-defined guidelines
- Mutation status classification
- Report creation in multiple languages



Microfluidic capillary electrophoresis analysis of a Coriell female normal sample (NA07538), and a Coriell male Fragile X sample (NA06852) with >200 repeats, together with the FraXsoft result interpretation and classification.

FRAGILEASE™ ASSAY CHARACTERISTICS

- Accurate amplification and detection of repeat sizes with > 900 repeats
- Precise repeat quantification of normal, premutation and full mutation samples
- Accurate detection of both alleles in female samples reducing the need for Southern blot reflexing
- High and low throughput options for cost- and time-efficient analyses
- 96 or 1152 reactions per kit
- Turnaround time from DNA to report approximately six hours
- CE marked for IVD use

WORKFLOW

1. FragilEase™ PCR
2. PCR Purification
3. LabChip MultiDX

ORDERING INFORMATION

Product Description	Part Number
FragilEase™ (96 reactions)	3101-0010
FragilEase™ (1152 reactions)	3101-001B

Product Description	Part Number
LabChip MultiDX	CLS136531
LabChip MultiDx 5K Chip (for 400 samples)	CLS760683
LabChip MultiDx 5K reagent Kit (for 4800 samples)	CLS760682

Products discussed in this brochure are not available in the USA and Canada.
In other countries please check availability with your PerkinElmer sales representative.

All PerkinElmer products may not be available in all countries.
For information on availability please contact your local representative.

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