ISO 13485

LabGscan™ FRAXA PCR Kit

“ **Fragile X syndrome** is a genetic hereditary condition that causes a scope of formative issues including learning handicaps and psychological debilitation. Generally, males are more seriously influenced by this disorder than females. Influenced individuals normally have delayed advancement of discourse and language by age 2. ”

LabGenomics

LabGscan™ FRAXA PCR Kit

Intended Use

The LabGscan™ FRAXA PCR kit is an in vitro diagnostic test based on PCR technology for the amplification and detection of CGG repeats in the 5'-untranslated region (5'-UTR) of FMR1 (Fragile X mental retardation 1) gene. The kit aids to diagnose 3 genotypes of normal, pre-mutation and full mutation(Table 1, Figure 1), and identify female carriers for fragile X syndrome.

Allele classification	CGG Repeats
Normal	Up to 44 repeats
Intermediate (Gray Zone)	45~54 repeats
Pre-mutation	55~200 repeats
Full mutation	Greater than 200 repeats

Table 1. Recommended allele classification

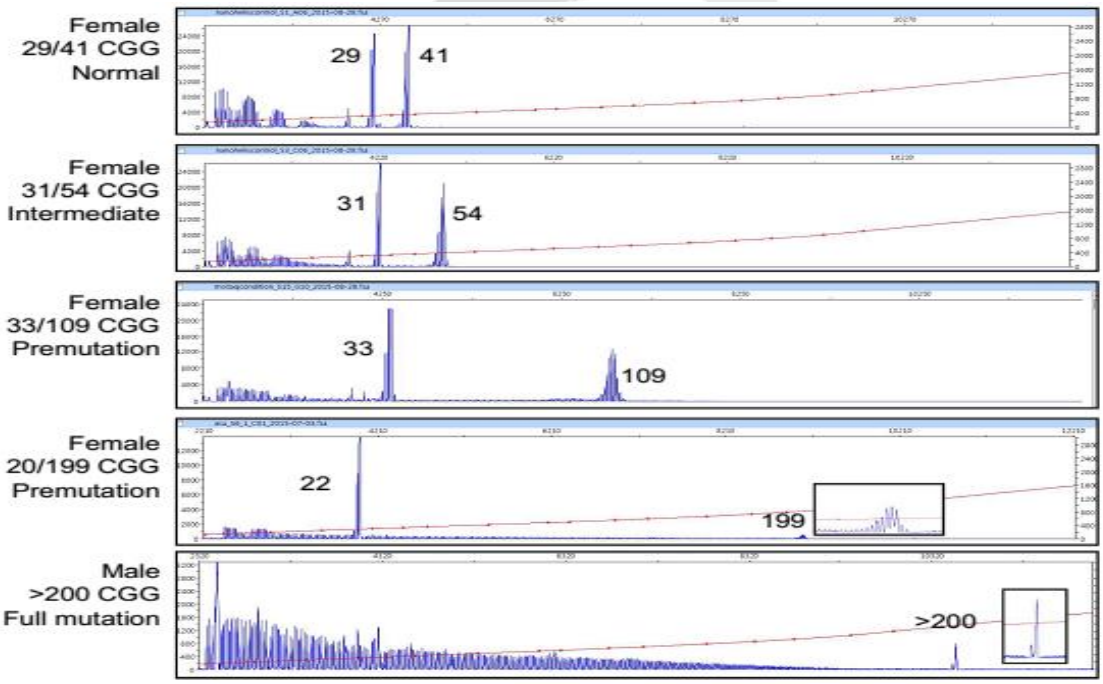

Figure 1. Examples of FRAXA genotyping

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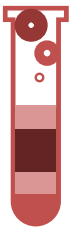
Screening Target and Purpose

Test Screens for possibility of Fragile X Syndrome infant. Fragile X Syndrome is a CGG Trinucleotide repetitive disorder of the FMR1.

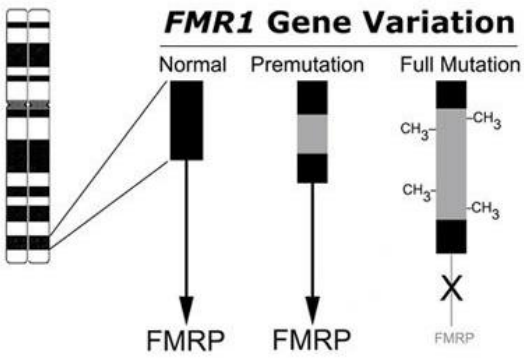


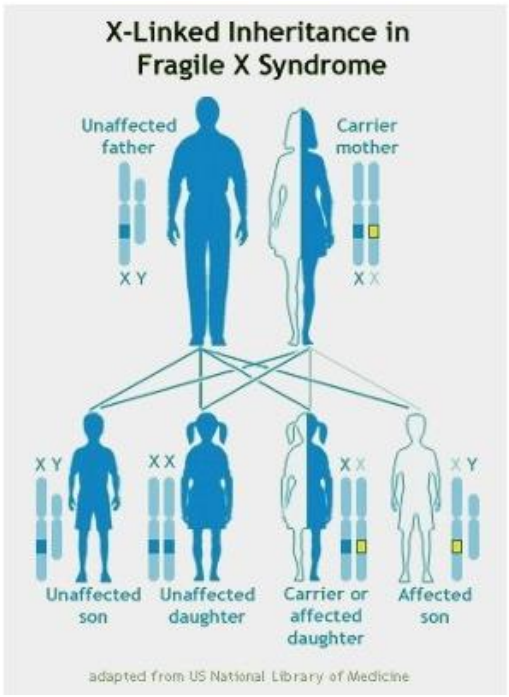
Utilizing test results

Detection through prenatal test is important for Fragile X Syndrome because unaffected mother can give birth to an affected infant, when X chromosome gene sequence are repeated only certain amounts, a state designed as 'carrier'.

Method of analysis

1st to perform prenatal screening test for Fragile X Syndrome since 2002. Developed novel molecular prenatal tests based on cutting-edge molecular biologic technology.





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LabGscan™ FRAXA PCR Kit

“ ”

The LabGscan™ FRAXA PCR Kit are designed for detection and identification of human genetic diseases through LabGenomic’s broad experience and high technology of the molecular genetics

Validity Period

12 months from the date of manufacture (open, unopened)

Storage Temperature

-20±5°C

Applied Sample

Genomic DNA extracted from whole blood (EDTA Whole Blood)

Reagents

Item No	Reagents	Presentation 25 reactions	Presentation 50 reactions	Presentation 100 reactions
1	FMR1 Primer Mix	1 vial, 25 μL	1 vial, 50 μL	1 vial, 100 μL
2	GC-Rich Amp Buffer	1 vial, 300 μL	1 vial, 600 μL	1 vial, 1.2 mL
3	Polymerase Mix	1 vial, 12.5 μL	1 vial, 25 μL	1 vial, 50 μL
4	ROX 1000 Size Ladder	1 vial, 50 μL	1 vial, 100 μL	1 vial, 200 μL

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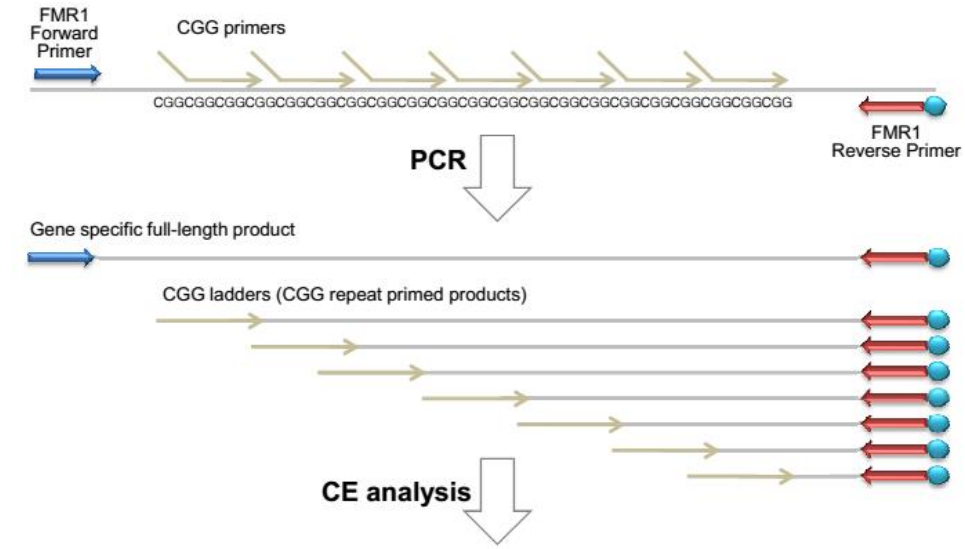
" TP-PCR (triplet repeat primed PCR) "



Sample Preparation

- Extraction of DNA from clinical samples (Whole blood etc.)

FMR1 PCR

- Target amplification
- Approximately 5.5 hours

CE Sizing

- Sizing the amplicons using Genetic Analyzer
- Approximately 1.5 hours

Results

- Converting base pairs to number of CGG repeats
- Approximately 0.5 hours

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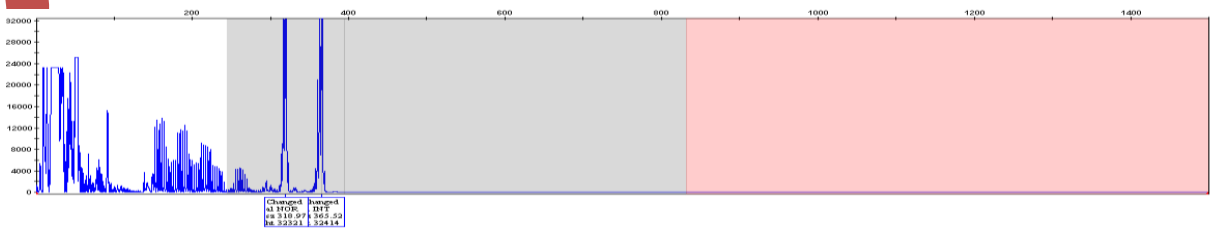
“ FRAXA PCR Kit Analyzing program ”

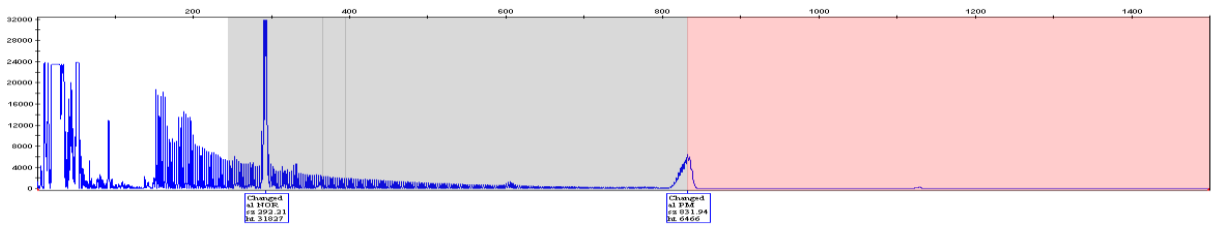
Elements to be set during GeneMapper analysis

- Analysis method
- Panel
- Size standard

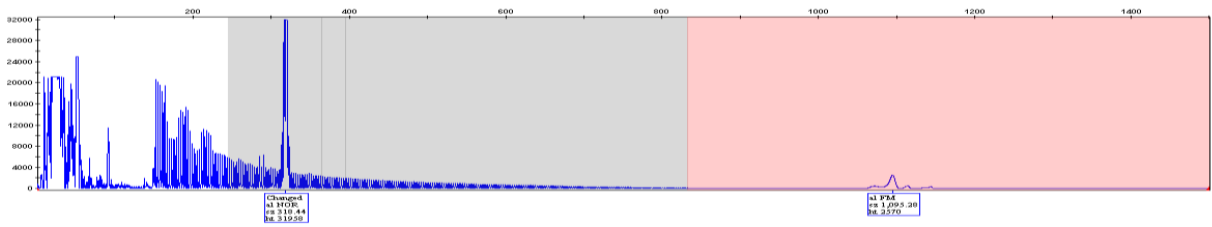
Female normal



Female Pre mutation



Female Full mutation



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PCR

Veriti
(Thermo Fisher Scientific)
C1000 & S1000 (Bio-Rad)

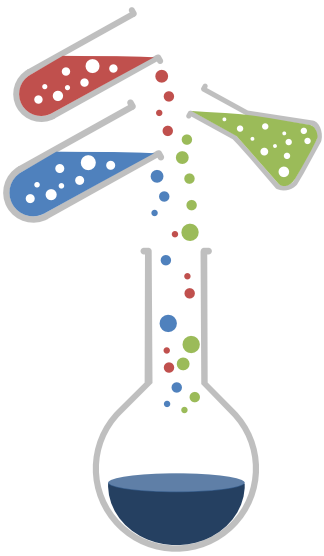
Recommended Instruments

Genetic Analyzer

3730, 3730xl DNA Analyzer (Applied Biosystems)
3130, 3130xl DNA Analyzer (Applied Biosystems)
3500, 3500xL Genetic Analyzer (Applied Biosystems)

Instrument	Capillary length	Injection	Run time
3730, 3730xl	**50cm**	**2.5kV, 20s**	**4000s**
3130, 31310xl	36cm	2.5kV, 20s	2400s
3500, 3500xL	50cm	2.5kV, 20s	4000s

Calculating CGG repeat



$$CGG_i = \frac{Peak_i - c_0}{m_0}$$

$Peak_i$ - size in base pairs of a given product peak

c_0 - size correction factor

m_0 - mobility correction factor for each CGG repeat

Configuration	c_0	m_0
3730, 3730xl 50cm	**231.6**	**2.995**
3130, 3130xl 36cm	229.5	2.97
3500, 3500xl 50cm	231.6	2.98

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