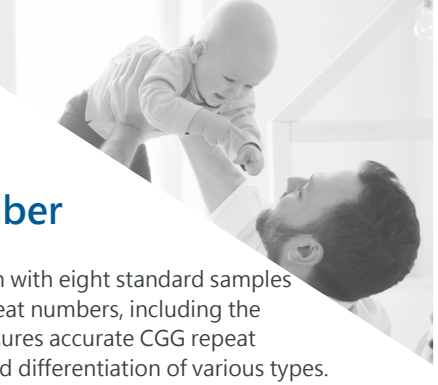
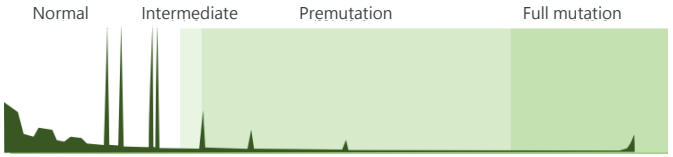
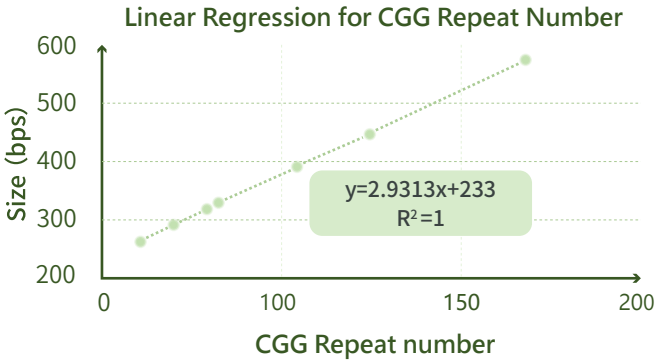


## Accurate Repeat Number

Using linear regression with eight standard samples covering different repeat numbers, including the positive threshold, ensures accurate CGG repeat number calculation and differentiation of various types.




## Experimental Procedure

After amplification, capillary electrophoresis is conducted, followed by automated analysis to generate reports.


## Product Advantages

### High Sensitivity
Accurate detection with as low as 5ng DNA input

### High Accuracy
Linear regression with 8 repeat numbers, including positive threshold

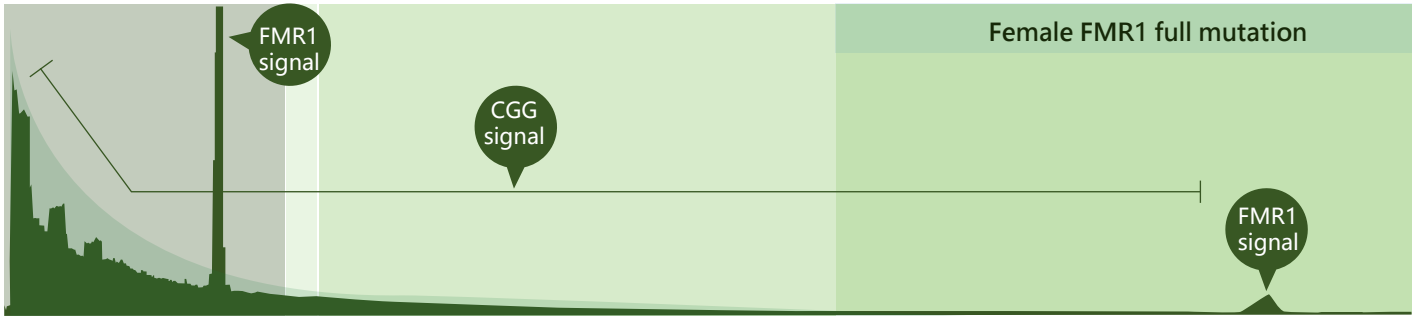
### Ultra-High Repeat Numbers
Can detect >1300 CGG repeats

### Prevention of False Negatives
CGG repeat number and FMR1 biallelic review

### Low Proportion Mosaic
Detects low proportion mosaic

### Fast Operation
Results obtained within 5 hours

## Experimental Results




rigin

# PCR-CE FMR1 Kit

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*Answers for better life*

# Fragile X Syndrome (FXS)

Fragile X Syndrome is the most common inherited intellectual disability disorder, which is an X-linked genetic condition with a carrier rate of 1/250.

Characterized by an abnormal increase in CGG repeats in the non-coding region of the FMR1 gene on the X chromosome, results in a deficiency of the FMRP protein, crucial for brain neuroconduction. Symptoms include intellectual disability, autism, developmental delay, seizures, and behavioral disorders.

## International guidelines

ACOG The American College of Obstetricians and Gynecologists

ACMG American College of Medical Genetics and Genomics

The American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG) recommend screening for fragile X syndrome in women of childbearing age to prevent the birth of affected children.

## Clinical Classification

Based on the number of CGG repeats, FMR1 genotypes can be classified into normal, intermediate, premutation, and full mutation types. Premutation carriers of fragile X syndrome can cause tremor/ataxia syndrome in males and premature ovarian insufficiency in females.

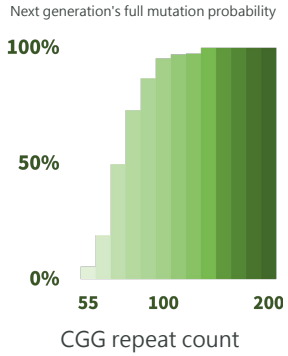
## Genetic Types and Clinical Manifestations

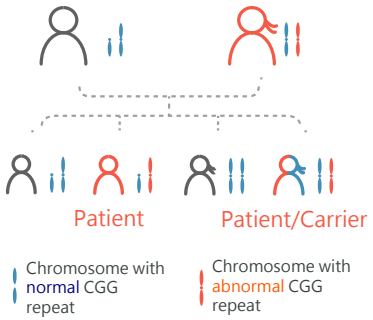
| CGG Repeats | | Genotypes | Male | Female | Recommendations |
|---|---|---|---|---|---|
| >200 | Prevalence 1/3600~1/5000 | Full mutation | Typical FXS Symptoms | 50% Intellectual Disability | Prenatal Diagnosis |
| 55-200 | 1/250 | Premutation | 40% FXTAS | 20% FXPOI | Prenatal Diagnosis |
| 45-54 | | Intermediate | Asymptomatic | | Newborn screening |
| <45 | | Normal | Asymptomatic | | - |

CGGCGGCGGCGGCGGCGG

## Inheritance Pattern

The prevalence of affected males is 1/3600, higher than females. FMR1 gene shows dynamic mutation. Higher CGG repeats in premutation females increase the chance of full mutation in offspring, with a 50% chance of having an affected child.




## Population for Use

- Children with unexplained intellectual disabilities, autism, or behavioral or language disorders
- Fetuses of Fragile X Syndrome carriers
- The following family history:
  - Fragile X Syndrome
  - Unexplained Intellectual Disabilities
  - Premature Ovarian Insufficiency
  - Tremor/Ataxia Syndrome
- Women with infertility, recurrent miscarriages
- Families preparing for conception and in need of fertility assistance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