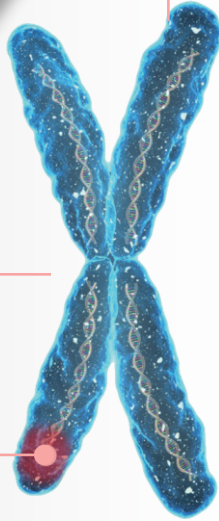


FMR1
q27.3



Obtained CE certification

Microreader™ Fragile X Syndrome Detection Kit

Fragile X Syndrome (Fragile X Syndrome) is an X-linked genetic disease and one of the most common inherited intellectual disability diseases, second only to Down syndrome in incidence. The cause of the disease is the extended mutation of the FMR1 gene (CGG)_n on the X chromosome, which leads to the lack of FMRP protein that maintains normal nerve conduction in the brain. Patients with the disease will have severe mental retardation, developmental delay, language barriers and behavioral problems, including ADHD and autism syndrome, inattention, and accompanied by epilepsy and other symptoms. The kit adopts the three-primer PCR technology recognized by international authoritative guidelines, which can accurately calculate the number of CGG repeats, quickly and accurately distinguish different genotype groups, diagnose patients with fragile X syndrome, and meet the different needs of clinical testing.

Features

Sensitive and accurate detection

Kit can amplify the full-length sequence of CGG repeats; CGG repeats greater than 200 can also be detected; the detection results are 100% reproducible

The perfect detection system

Kit can accurately distinguish various genotypes, accurately determine female homozygotes and heterozygotes, and effectively avoid false negatives

Simple and fast operation

The whole process only takes around 6 hours, easy to operate, suitable for automation and batch testing

Rich project experience

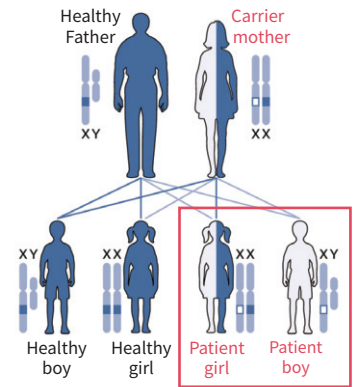
With many years of clinical testing experience, the kit was used for providing services in many testing centers worldwide, and related data was published in international journals ^[1]

- Women of childbearing age
- Infertile women
- Fetuses of fragile X carriers
- People with a family history of Fragile X Syndrome
- Children with unexplained mental retardation, autism, behavioral disorders, and language disorders
- People with a family history of unexplained mental retardation
- People with premature ovarian failure and their family history
- People with tremor-ataxia and their family history
- Women with unexplained abortions and recurrent miscarriages

Genetic pattern

Because men have only one X chromosome, they are more susceptible to the disease, about one in 4,000 people have this disease; the incidence rate of women is relatively low, but the pre-mutation carrier rate of Chinese women is about 1/634^[2], and there are dynamic mutations, that is, women carriers are more likely to give birth to children with full mutations.

According to the national Fragile X foundation (National Fragile X Foundation) in America, the probability of a female carrier to give birth to a child with the disease can be as high as ½ ! Therefore, it is particularly important to carry out prenatal or pre-pregnancy FRM1 gene detection for women of childbearing age, and then guide fertility.



credit: U.S. National Library of Medicine

Description of FMR1 gene mutation types and clinical manifestations^[3]

CGG repeat count genotype	Class	Clinical manifestations		Suggestion
		Female	Male	
<45	Normal	-	-	-
45-54	Intermediate	-	-	Recommended for newborns FMR1 gene mutation screening
55-200	Pre-mutation	About 20% suffer from premature ovarian failure	About 40% of adult males suffer from Tremor and ataxia syndrome	Prenatal diagnosis is recommended to confirm whether the fetus is a patient with fragile X syndrome
>200	Full-mutation	30-40% have fragile X syndrome	100% of patients with fragile X syndrome	

Detection process

1 DNA extraction



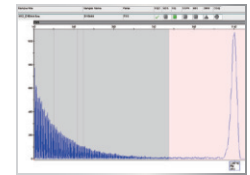
2 Fluorescent PCR amplification



3 Detection on Capillary electrophoresis platform



4 Results interpretation



Sampling requirements

2-4 ml EDTA anticoagulant blood, ≥50mg chorion sample, ≥100mg tissue sample ► transported and stored at 2-4°C
 2 cm² blood stored on FTA blood card ► transported and stored at room temperature

Applicable instrument

PCR instrument: Life Technologies Holdings Pte Ltd: Veriti, Veriti Dx, 9700

Genetic analyzer: Life Technologies Holdings Pte Ltd: 3500 Dx, 3500 xL Dx; Sequstudio

*References : [1] Chen X, et al. BMC pediatr. 2015 jul 15;15:77.

[2] Gao F, et al. Molecular genetics & genomic medicine.

[3] Hagerman R, et al. Pediatrics. 2009 jan;123(1):378-90.

* This product is for research use.

* This information is only for reference by relevant medical professionals. Please refer to the instruction manual for contraindications or precautions.

