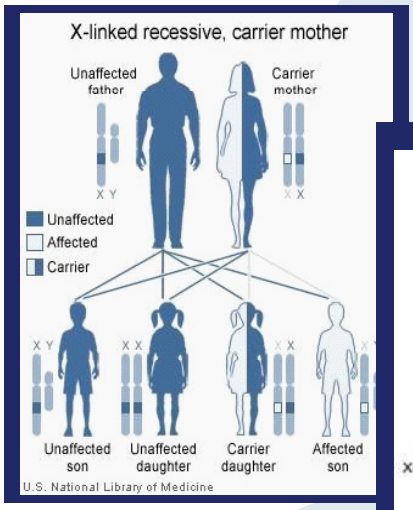
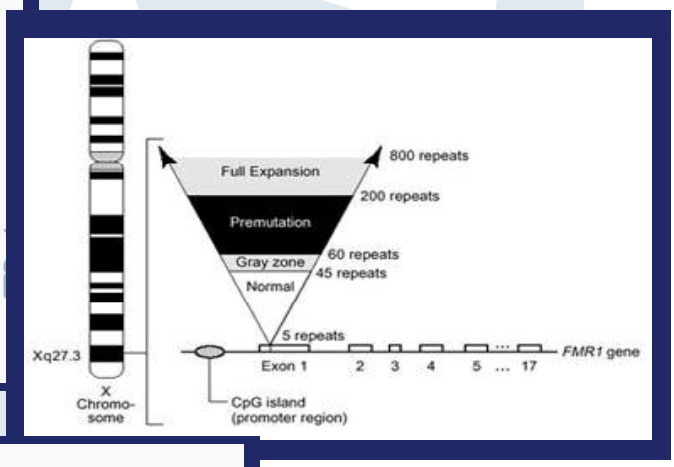
CYTOGEN

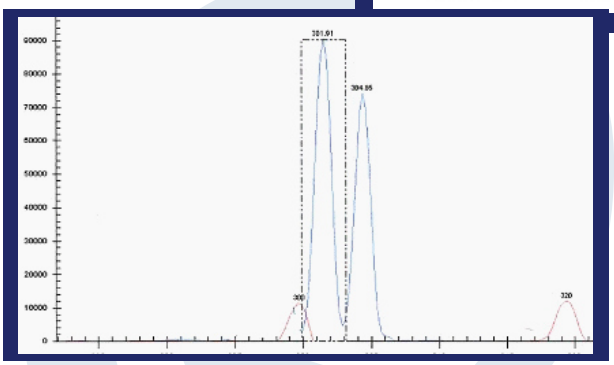
Produkte für Medizin und Forschung GmbH

FRAXA 1 KIT-FL

System for the molecular analysis of the Fragile X Syndrome by automated capillary DNA sequencer







EXPERTEAM

l'esperienza in biologia molecolare

CE IVD

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The fragile X syndrome (Martin-Bell syndrome) is the most common cause of inherited mental retardation. In affected boys, delay in language acquisition and/or behavioural problems are often the presenting symptoms. In addition to cognitive deficits, the phenotype of fragile X syndrome includes mild dysmporphic features and macroorchidism.
Behavioural disturbances including attention-deficit hyperactivity or autistic- like behaviuor can be observed. It is caused in most cases by **expansions of a (CGG) trinucleotide repeat** in the 5'UTR region of the FMR1 gene and subsequent abnormal methylation of neighbouring CpG island leading to the loss of the protein product FMRP.
In normal subjects the CGG repeat is polymorphic and contains from **6** to about **50** trinucleotide repeats; the most common alleles show 29-30 repeats. Alleles in the **59-200** repeat range without abnormal metilation are referred to as **premutations**; they are unstable and, when transmitted by a female, have a risk of expanding to a full mutation. Alleles with CGG repeats in the intermediate range (gray zone) between normal and premutation (**51-58** repeats) can show the same instability upon maternal and paternal transmission.
The **full mutation** that causes the disease is a large expansion exceeding **200** repeats and is usually accompanied by methylation of the adjacent promoter region which results in transcriptional silencing of FMR1.

Containing of the kit

Label	Contents
6FAM/Cy5 FRAXA MIX	Mix for the amplification of the FMR-1 gene
6FAM/Cy5 AMXY MIX	Mix for the amplification of the control gene
C WT	Wild-type Control
P DNA polymerase	DNA polymerase with "proof- reading" 3'-5' activity
ExperTaq polymerase	Taq DNA polymerase

How does the kit work?

The Fraxa 1 kit-FL is a system for the amplification of the FMR1 (CGG)n repeat region thank you to the use of a particular **P DNA polymerase**.
This enzyme possesses proofreading 3´ to 5´exonuclease activity and its fidelity, accuracy, and specificity make it ideal for the amplification of GC-rich templates, especially the premutated alleles (100-120 repeats). It does not amplified longer DNA sequences presenting the full mutation (CGG > 200).
A master mix for a **control gene** amplification is provided to avoid false results. This control allow also to verified the sex of the sample analyzed. After DNA amplification, PCR products are separated by **capillary electrophoresis**. The result interpretation is based on the absence/presence and on the determination of the lenght of the CGG repeat in the FMR1 gene.

When to use this kit?

This kit is used as simple and rapid pre- screening test to exclude X-fragile diagnosis in most of the case when examination is requested.
Permit to avoid to proceed with the Southern blot, doing this analysis only on samples which fail to amplify (males, FMR-1 gene) or when a female sample show a single allele.

Starting samples: peripheral blood. **DNA isolation method:** QIAamp DNA blood kit, QIAcube, QIAsymphony (Qiagen), High Pure PCR template preparation kit (Roche). **DNA Sequencer:** CEQ 8000/8800 Genetic Analysis System (Beckman Coulter); 310, 3100, 3130, 3730, 3500 Genetic Analyzers (Applied Biosytems).

Product	Unit	Cat.-No.
FraxA 1 Kit - FL	40 tests	FL FR.01FL

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