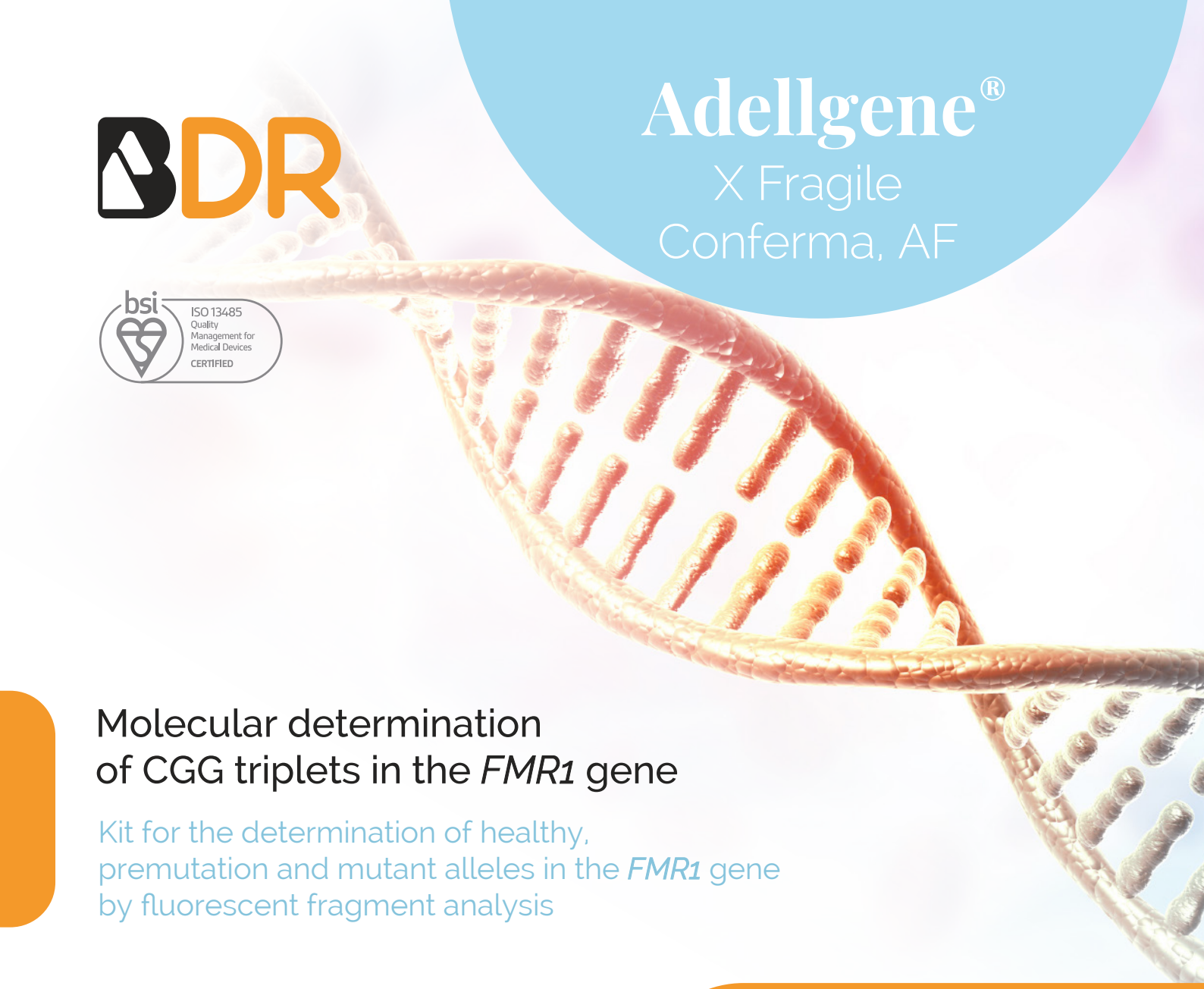
Adellgene®
X Fragile
Conferma, AF



Molecular determination
of CGG triplets in the *FMR1* gene

Kit for the determination of healthy,
premutation and mutant alleles in the *FMR1* gene
by fluorescent fragment analysis

About
Adellgene® X Fragile – Conferma, AF

Fragile X Syndrome (FXS, OMIM #300624) is an **X-linked disease** that is primarily based on the genomic expansion of a triplet of nucleotides (CGG), and aberrant methylation of the promoter region.

FXS has a prevalence of 1 in **4000 males** and 1 in **8000 females**, and is associated with mental retardation, autism, and mental and emotional changes. Affected individuals show a striking phenotype consisting on large ears and a prominent jaw.

Depending on the number of repetitions of this triplet, **four categories** can be established:
- Less than 45 repeats: individuals with **healthy alleles**.
- From 45 to 54 repeats: individuals with **intermediate alleles**.
- From 55 to 200 repeats: individuals with **premutation alleles**.
- Over 200 repeats: individuals with **mutant alleles.**

Intended use

Adellgene® Fragile X is a semi-automated *in vitro* diagnostic kit for use in clinical laboratories allowing quantitative determination of the number of CGG triplet repeats in the 5' untranslated region of gene for fragile X mental retardation ("Fragile X mental retardation-1": *FMR1*; or FXS), as an aid in the clinical diagnosis of disease associated with Fragile X syndrome, fragile X-associated tremor/ataxia syndrome (FXTAS), and fragile X-associated primary ovarian insufficiency (FXPOI).

The kit allows the quantification of the size of healthy, intermediate, premutant and expanded alleles with a size equal to or less than 200 repeats. The expansions of more than 200 repeats can be detected, but not quantified.

The procedure is based on the amplification of genomic DNA, extracted from whole blood and/or buccal swab, by Triplet Repeat Primed polymerase chain reaction (TP-PCR) with fluorescent primers, subsequent analysis of the size of the amplified fluorescent fragments in a capillary sequencer and conversion of the fragment size into its corresponding number of repeats.

The patient referred by the corresponding health specialist (e.g., neurologist) may be subject to this determination, taking into account the compatibility of the symptoms presented (mainly for FXS: developmental delay, intellectual disability, characteristic craniofacial features, autism spectrum disorder, hyperactive and/or impulsive behaviour; for FXTAS: late-onset, progressive cerebellar ataxia and/or intention tremor; and for FXPOI: hypergonadotropic hypogonadism before age 40 years), and/or family history.

The intended user of this kit is technical personnel trained and qualified to perform the protocol described in the instructions for use and interpretation of its results.

Workflow

Product Information

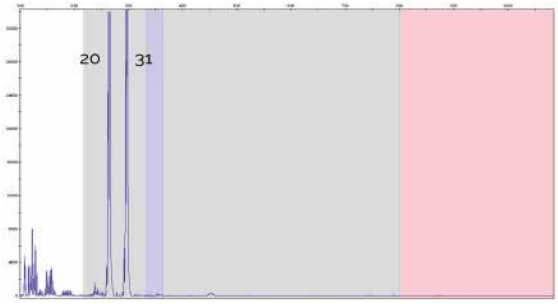
25 tests	- CODE: AA1603/25	UDI-DI: 8437016942468
100 tests	- CODE: AA1603/100	UDI-DI: 8437016942475

DESCRIPTION: Adellgene® X Fragile - Conferma, AF

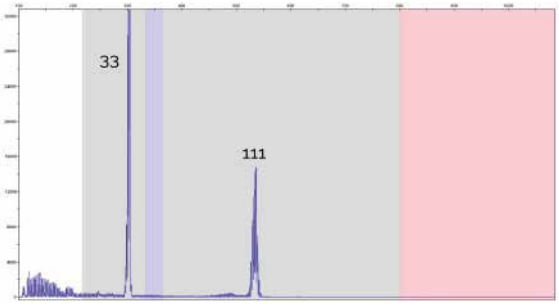
Notified body 2797 CE-IVD certified

Results

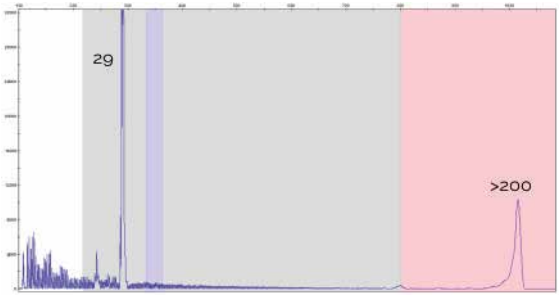
Healthy heterozygous female sample



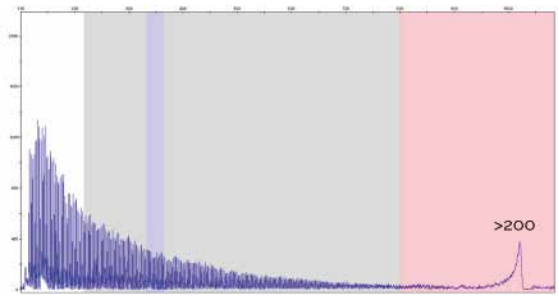
Premutation heterozygous female sample



Full expanded female sample



Full expanded male sample



Limitations

-The kit can detect alleles with a number of repeats from 5 and quantify up to 200.
-This kit does not allow accurate determination of the number and location of AGG interruptions. Additional analyses are recommended for the resolution of these alleles.
-The presence of variants in the regions where the primers hybridise may result in the lack of definition of an allele or an abrupt drop in intensity. Other technologies may be necessary for the resolution of such alleles.
-The interpretation of the results obtained must always be supervised by qualified personnel.

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