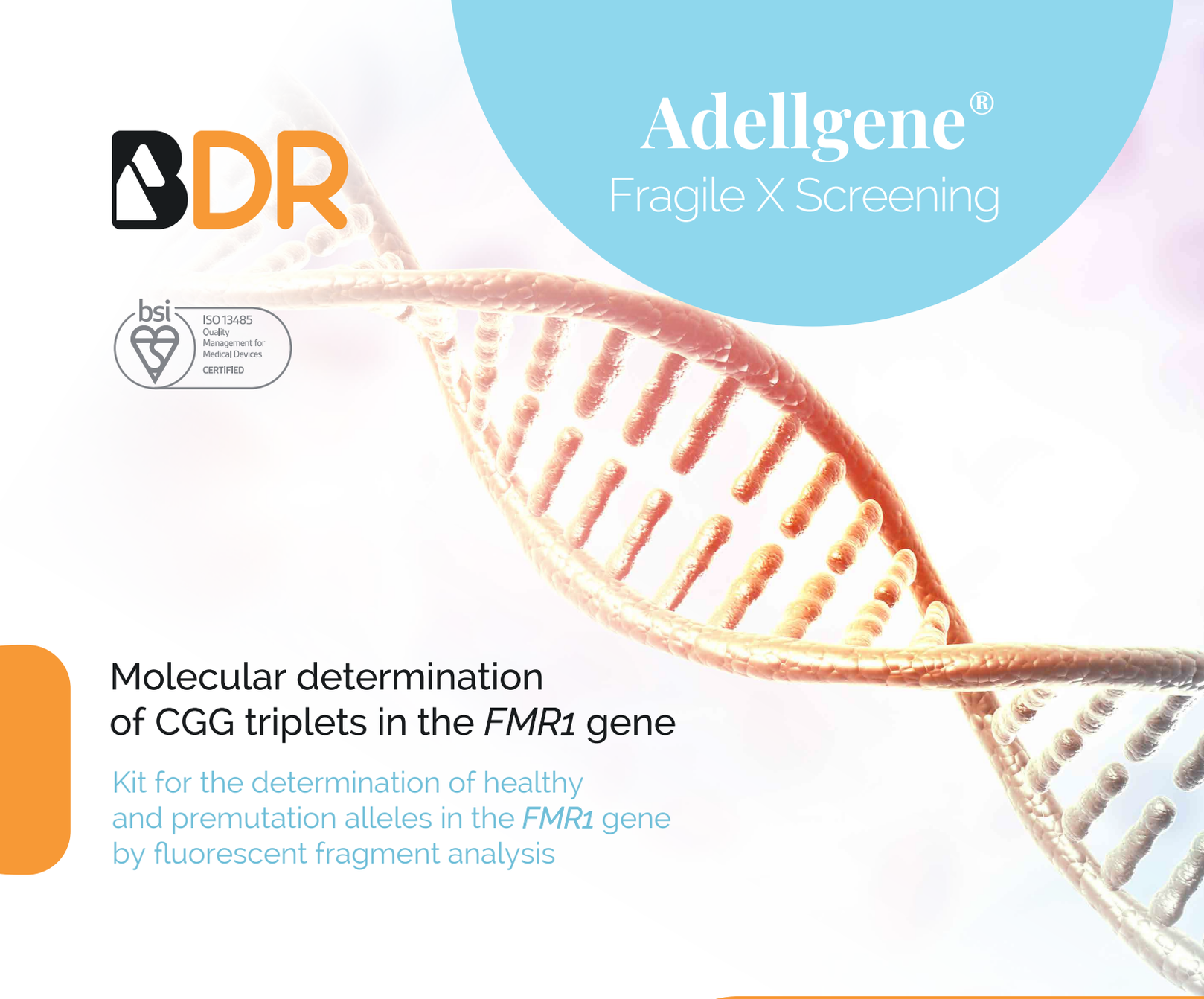
# Adellgene®
## Fragile X Screening



ISO 13485 Quality Management for Medical Devices CERTIFIED

## Molecular determination of CGG triplets in the *FMR1* gene

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

# About Adellgene® Fragile X Screening

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**. Affected individuals show a striking phenotype consisting on large ears and a prominent jaw.

Depending on the number of repetitions of this triplet, **three categories** can be established:
- Up to 45 or 55 repeats: individuals with **healthy alleles**.
- From 45 or 55 to 200 repeats: individuals with **premutation alleles**.
- Over 200 repeats: individuals with **mutant alleles**.

# Intended use

Adellgene® Fragile X Screening is a semi-automated *in vitro* diagnostic kit designed for use in clinical laboratories, which quantitatively determines the number of CGG triplet repeats (cytosine-guanine-guanine) in the 5' untranslated region of the gene for fragile X mental retardation ("Fragile X mental retardation-1": *FMR1*). It aims to aid diagnosis of the clinical disease associated with Fragile X syndrome, for example, mental retardation, primary ovarian failure, and tremors / ataxia.

This kit can be used for the determination of the number of repeats present in healthy individuals (up to 45/55 CGG repeats) and premutated individuals (from 45/55 to 200 CGG triplet repeats). Female samples reporting only one peak in the results' electropherogram and male samples with no peak must be analyzed using other appropriate technique, for example, TP-PCR, in order to verify the non-existence of a fully mutated allele (>200 CGG repeats).

The technology is based on the polymerase chain reaction (PCR) amplification of genomic DNA extracted from peripheral blood, followed by fluorescence analysis of the size of the PCR fragments obtained by a genetic analyzer and conversion of that size into the respective number of CGG repeats.

Patients who can benefit from this determination are those referred by a specialist. The intended user of the kit is technical personnel trained to carry out the protocol and the interpretation of results described in the Instructions for Use.

# Workflow

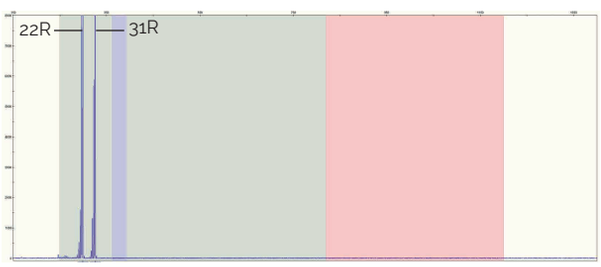

# Product Information

16 tests - CODE: **AD-FX-16** UDI-DI: **8437016942291**
48 tests - CODE: **AD-FX-48** UDI-DI: **8437016942307**

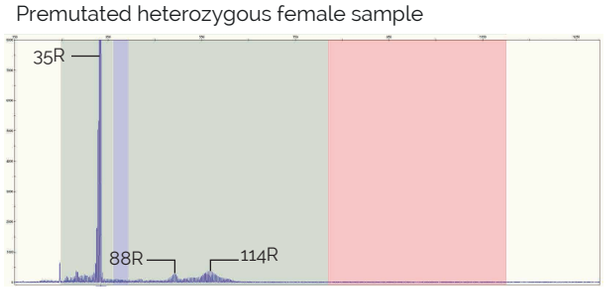
DESCRIPTION: **Adellgene® Fragile X Screening**
CE-IVD certified

# Results

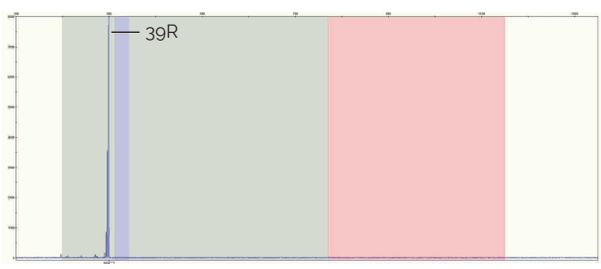
Healthy heterozygous female sample



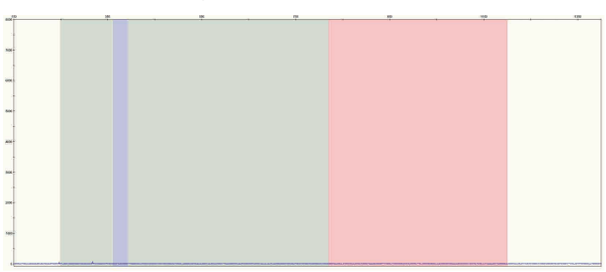

Premutated heterozygous female sample




Mutated female sample




Mutated male sample



# Limitations

- Mutations (point mutations, insertions, deletions) at amplification primer sites are possible and may result in the lack of allele definition. Other technologies could be necessary to resolve the genotyping.
- Homozygous results and males with no allele detected, must be confirmed by alternative procedures.
- Data and result interpretation should be revised by qualified personnel.