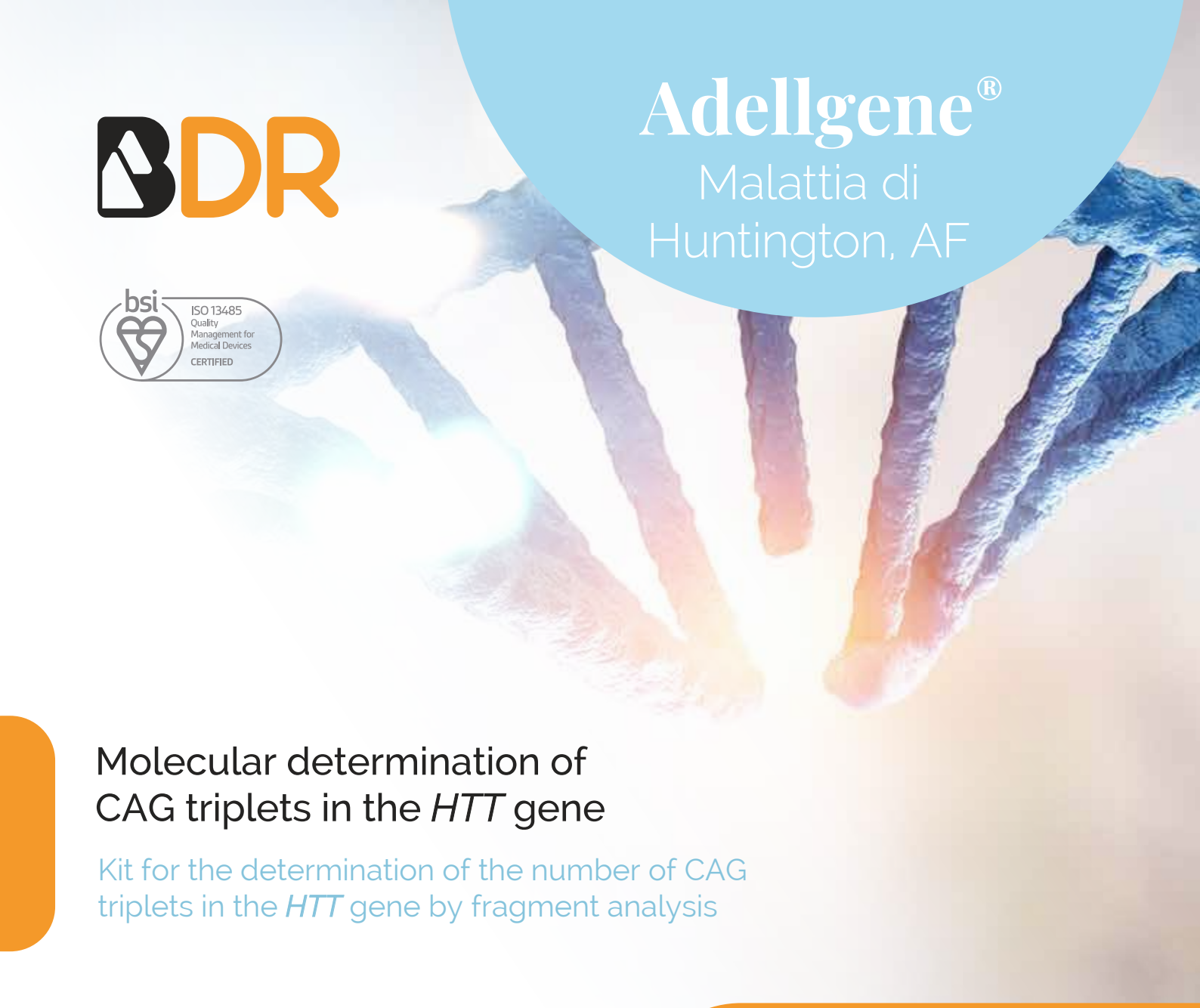
Adellgene®
Malattia di Huntington, AF



Molecular determination of CAG triplets in the *HTT* gene

Kit for the determination of the number of CAG triplets in the *HTT* gene by fragment analysis

About Adellgene® Malattia di Huntington, AF

Huntington's disease (HD) is a hereditary neurodegenerative disorder that affects the central nervous system. Symptoms typically appear between the ages of 30 and 50, with a global prevalence ranging from 5 to 10 cases per 100,000 people, and a higher incidence in populations of European descent.

The genetic basis of Huntington's disease lies in a pathological expansion of CAG trinucleotide repeats in the *HTT* gene, located on chromosome 4p16.3.4 The CAG trinucleotide expansion leads to the production of a mutated huntingtin protein. This structural modification alters the protein's conformation, promoting its aggregation and accumulation in both the cytoplasm and nucleus of neurons. These aggregates interfere with cellular function, energy metabolism, and calcium homeostasis, ultimately leading to neuronal death.

Why Adellgene®?

- **CE- IVDR** certification
- A **single reaction** for **quantification** of healthy, intermediate, reduced and full penetrance alleles up to 200 CAG repeats and **detection** of expansions of more than 200 repeats.
- **16 tests format** specially adapted to laboratory routines.
- **Upgraded content:** Positive and Negative controls and RED1000 Size Ladder included.
- **Open system:** validated in various capillary sequencers; 3130xl, SeqStudio, 3500xl and Spectrum Compact.

Workflow

Product Information

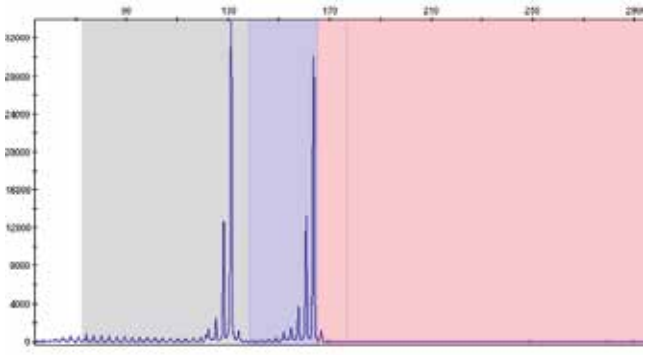
Adellgene® Malattia di Huntington, AF

CODE: AA1611/16 / UDI-DI: 8437016942369

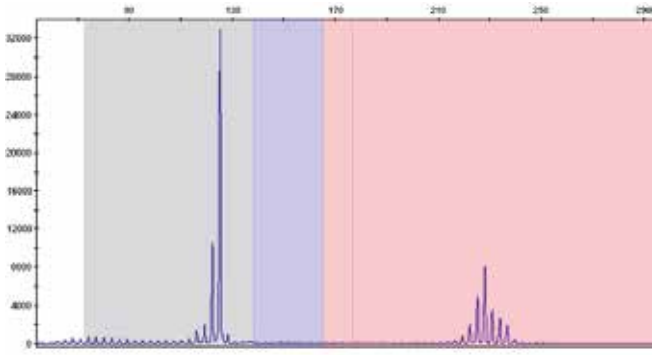
CE-IVDR Certified (Notified body 2797)

Results Interpretation

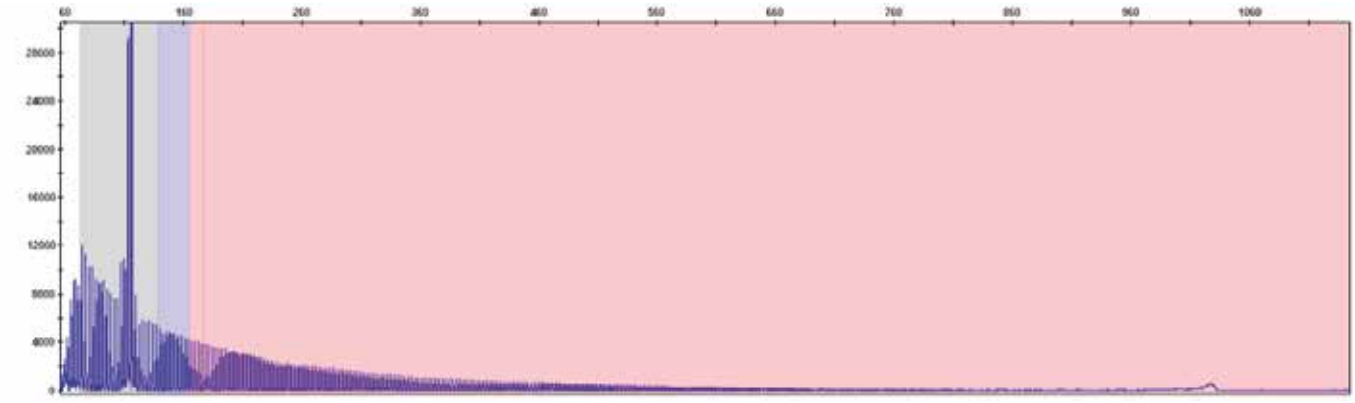
Sample with one normal allele and one intermediate allele.



Sample with one normal allele and one fully penetrant allele (<200 repeats).



Sample with one normal allele and one fully penetrant allele (>200 repeats).



Limitations

- The kit can detect alleles with a number of repeats from 5 and quantify up to 200.
- The presence of variants (point mutations, insertions, deletions) 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.

BDR
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