# Adellgene®
## Distrofia Miotonica
## Conferma, AF

ISO 13485 Quality Management for Medical Devices CERTIFIED

## Molecular determination of CTG triplets in the *DMPK* gene

Kit for the determination of the number of CTG triplets in the *DMPK* gene by fragment analysis

# About Adellgene® Distrofia Miotonica - Conferma, AF

Myotonic dystrophy type 1 (DM1), also known as Steinert disease, is an inherited neuromuscular disorder characterized by progressive muscle weakness and atrophy.

DM1 can manifest at any stage of life, from birth to adulthood. With a global prevalence of approximately 1 in 8,000, DM1 is the most common type of muscular dystrophy with adult onset.

DM1 is an autosomal dominant disorder caused by an abnormal expansion of CTG trinucleotide repeats in the 3' UTR region of the *DMPK* (Dystrophia Myotonica Protein Kinase) gene located on chromosome 19. The CTG repeat expansion affects the production and function of the DMPK protein and other cellular processes, leading to the diverse symptoms observed in DM1.

# Why Adellgene®?

- **CE- IVDR** certification
- A **single reaction** for **quantification** of healthy, premutation and expanded alleles up to 200 CTG 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

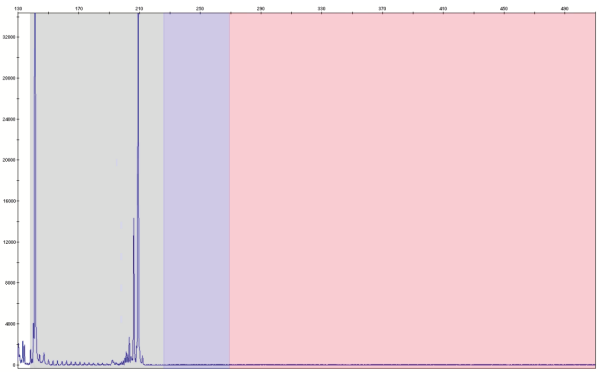
Adellgene® Distrofia Miotonica - Conferma, AF

CODE: AA1614/16 / UDI-DI: 8437016942345

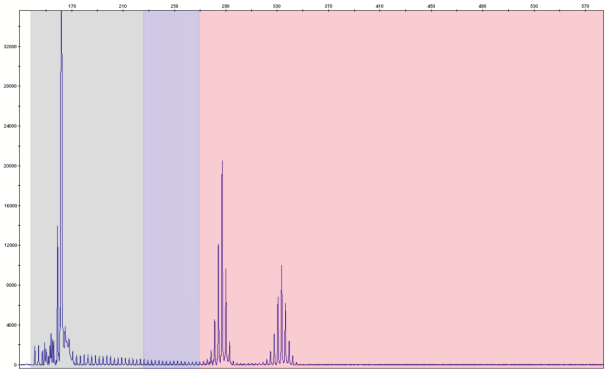
CE-IVDR Certified (Notified body 2797)

## Results Interpretation

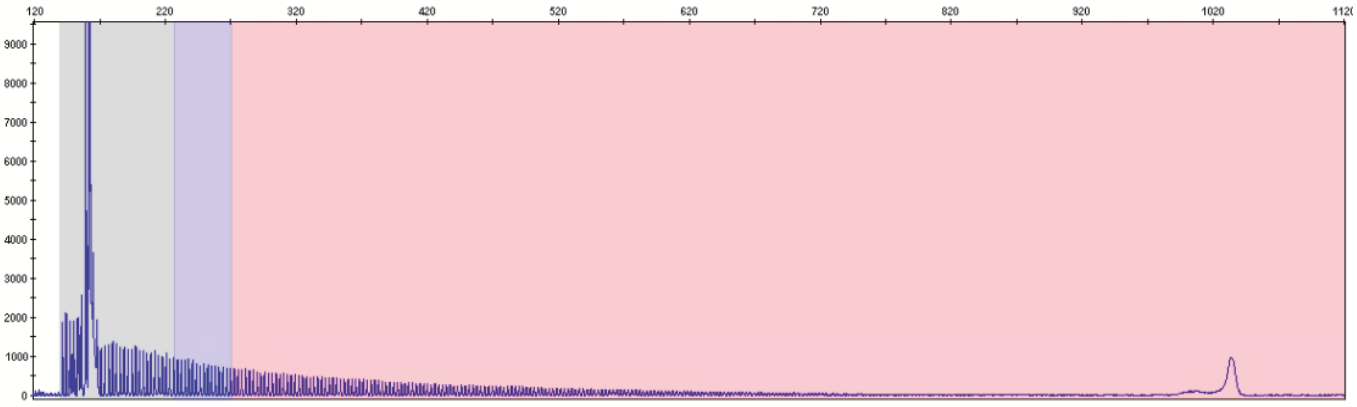
Sample with two wild-type alleles



Sample with one normal allele and one expanded allele (<200 repeats), showing somatic mosaicism



Sample with one normal allele and one expanded 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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