Adellgene®
Atassia di Friedreich, AF

Molecular determination of GAA triplets in the *FXN* gene

Kit for the determination of the number of GAA triplets in the *FXN* gene by fragment analysis

About Adellgene® Atassia di Friedreich, AF

Friedreich's ataxia (FA) is a rare, inherited neurodegenerative disorder that primarily affects the nervous system and muscles.

FA is the most common hereditary ataxia, with prevalence varying significantly across regions, ranging from approximately 1 in 20,000 in southwestern Europe to around 1 in 250,000 in northern and eastern Europe.

The disease is caused by variants in the *FXN* gene located on chromosome 9. Approximately 96% of individuals with FA have biallelic GAA trinucleotide repeat expansions in intron 1 of the *FXN* gene, while approximately 4% are compound heterozygotes, carrying an expanded GAA repeat in one allele and either an intragenic *FXN* pathogenic variant or a large deletion in the other allele. These genetic variants lead to reduced levels of frataxin protein, impairing mitochondrial function and resulting in oxidative stress and neurodegeneration.

Why Adellgene®?

- **CE- IVDR** certification
- A **single reaction** for **quantification** of healthy, intermediate and pathogenic 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

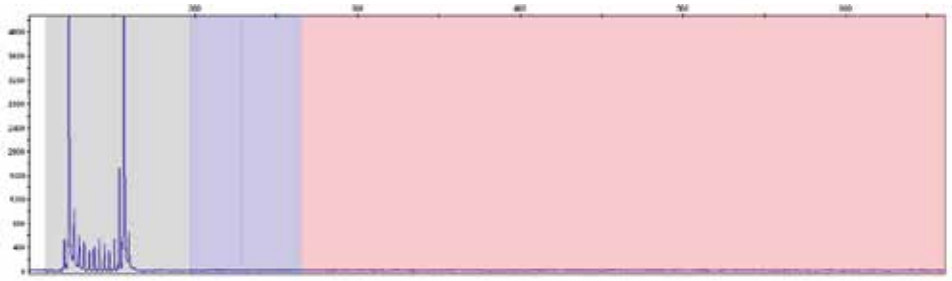
Adellgene® Atassia di Friedreich,AF

CODE: AA1615/16 / UDI-DI: 8437016942352

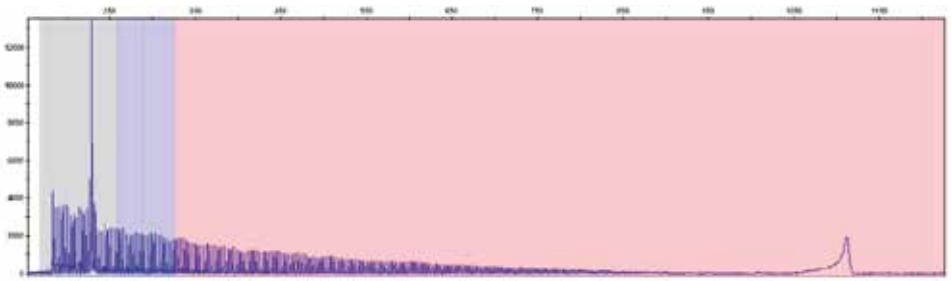
CE-IVDR Certified (Notified body 2797)

Results Interpretation

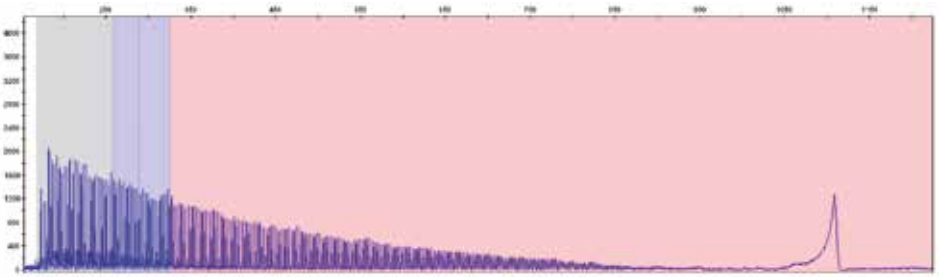
Sample with two wild-type alleles



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



Sample with two expanded alleles (>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
BLACKHILLS DIAGNOSTIC RESOURCES S.L.U.
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EXCLUSIVE DISTRIBUTOR
Nuclear Laser Medicine srl
Viale delle Industrie, 3 - 20049 Settala (Milan), ITALY
Phone +39 02 952451 - Fax +39 02 95245237-8
VAT 08763060152
segreteria@nlm.it - **www.nlm.it**