



Adellgene®

SCAs

Molecular determination of CAG triplets in the *SCA1*, *2*, *3*, *6*, *7*, *12*, *17* and *DRPLA* genes

Kit for the determination of the number of triplet repeats in the genes *ATXN1* (*SCA1*), *ATXN2* (*SCA2*), *ATXN3* (*SCA3*), *CACNA1A* (*SCA6*), *ATXN7* (*SCA7*), *PPP2R2B* (*SCA12*), *TBP* (*SCA17*), and *ATN1* (*DRPLA*), by fragment analysis.

About Adellgene® SCAs Kit

Spinocerebellar ataxias (SCAs) are a heterogeneous group of hereditary neurodegenerative disorders that mainly affect the cerebellum, leading to progressive loss of coordination and balance. These diseases are autosomal dominant and are characterized by a wide clinical and genetic variability.

At least twelve types of SCAs are caused by expansions of repetitions, among which six result from mutations caused by the expansion of the CAG trinucleotide which encodes polyglutamine stretches in proteins associated with these diseases: *SCA1*, *SCA2*, *SCA3*/Machado-Joseph disease, *SCA6*, *SCA7*, and *SCA17*. *SCA12* is also caused by the expansion of the CAG trinucleotide, although the mutation is found in the 5' UTR region of the gene. Additionally, another disorder related to a translated repetition of the CAG trinucleotide, dentatorubral-pallidoluysian atrophy (*DRPLA*), is classified as an SCA due to its clinical characteristics.

Intended use

Adellgene® is a semi-automated *in vitro* diagnostic kit for use in clinical laboratories allowing quantitative determination of the number of triplet repeats in genes *ATXN1* (SCA1), *ATXN2* (SCA2), *ATXN3* (SCA3), *CACNA1A* (SCA6), *ATXN7* (AAS7), *PPP2R2B* (SCA12), *TBP* (SCA17) and *ATN1* (DRPLA), as an aid in the clinical diagnosis of autosomal dominant spinocerebellar ataxias and dentatorubral-pallidoluysian atrophy (DRPLA).

The kit allows the quantification of the size of healthy, intermediate, uncertain and expanded alleles with a size equal to or less than 200 repeats. The expansions of more than 200 repeats described for SCA2 and SCA7 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 polymerase chain reaction (PCR or 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 (progressive cerebellar ataxia with motor abnormalities, dysarthria, ophthalmoplegia, involuntary movements/tremors, peripheral neuropathy and cognitive impairment), 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

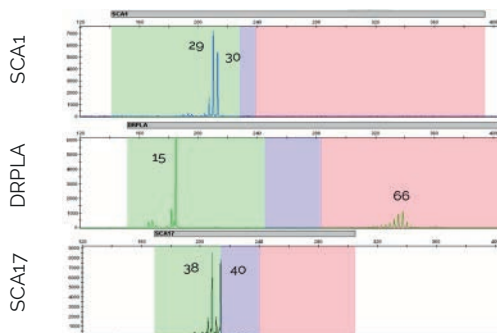
16 tests - CODE: AD-SCA-16 UDI-DI: 8437016942437

DESCRIPTION: Adellgene® SCAs 16

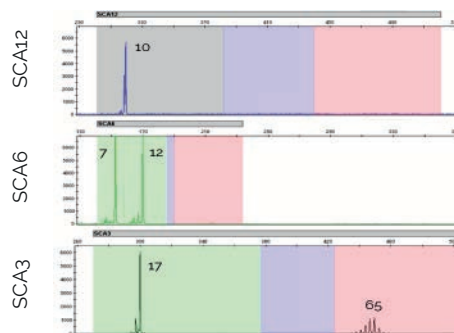
CE-IVDR certified

Results

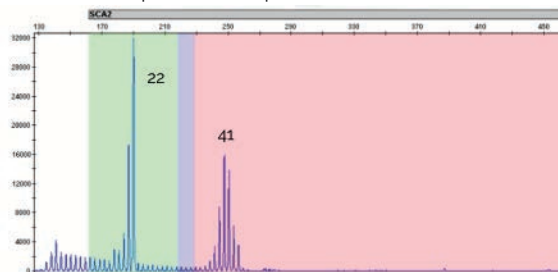
Reaction 1: Expanded sample for DRPLA. Healthy for SCA1 and SCA17



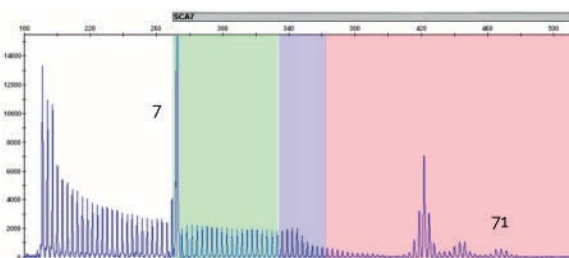
Reaction 2: Expanded sample for SCA3. Healthy for SCA6 and SCA12



Reaction 3: Expanded sample for SCA2



Reaction 4: Expanded sample for SCA7



Limitations

- This kit can quantify all alleles up to 200 CAG repeats.
- 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.
- Data and result interpretation should be revised by qualified personnel.