

# Methods to the diagnosis and prognosis of Machado-Joseph Disease

## KEYWORDS

Oligonucleotides; Polyglutamine Disorders; PCR-based diagnosis; CAG repeats; Machado-Joseph Disease/Spinocerebellar Ataxia type 3 (MJD/SCA3)

## TECHNOLOGY DESCRIPTION

Optimized oligonucleotide sequences for PCR-based diagnostic of the genetic etiology of Machado-Joseph Disorder (MJD), the CAG trinucleotide expansion within *ATXN3* gene: a) length of the expansion and b) single-nucleotide polymorphisms (SNPs) associated with the mutant allele. This method will allow the diagnosis and progression prediction of this disorder, as well as the design of specific gene therapy strategies.

Optimized oligonucleotides and probes to measure expression of mutant mRNA in Machado-Joseph Disorder to complement the genetic diagnosis and to evaluate the potential of gene-based therapies such as gene editing or silencing.

### Advantages:

In the detection of the number of CAG repeats:

- Easier and faster procedure to accurately diagnose of MJD

In the detection of SNPs associated with the CAG expansion:

- Identification of patients suitable for novel and allele-specific therapeutic strategies

In the measurement of mutant mRNA:

- To complement the genetic diagnosis
- Evaluation of the potential of gene-based therapies
- Evaluation of disease progression in disease models

**Faster, easier and more accurate diagnosis and prognosis prediction of Machado-Joseph Disease.**

## ADVANTAGES OVER ALTERNATIVE TECHNOLOGIES

PCR amplified products using polyacrylamide gel electrophoresis (PAGE):

- Less labour-intensive and time-consuming procedure
- More accurate and easier detection of the number of CAG repeats

Next-generation sequencing:

- Lower cost
- Feasible detection of the number of CAG repeats

## APPLICATIONS

Machado-Joseph Disease and other polyglutamine disorders:

- Diagnosis and progression prediction
- Improvement of molecular diagnosis
- Appropriate use and development of novel and allele-specific therapeutic strategies

## PATENT SPECIFICATIONS

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