Technology Offer

TARGETED AND MINIMAL INVASIVE GENE THERAPY FOR MACHADO-JOSEPH DISEASE

KEYWORDS

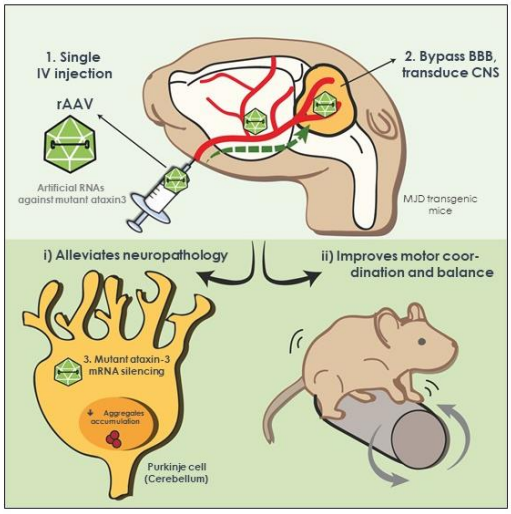
Machado-Joseph Disease, Spinocerebellar Ataxia 3 (SCA 3), Gene Therapy, RNA silencing, Adeno-associated viral vector (AAV)

TECHNOLOGY DESCRIPTION

This technology utilizes RNA silencing techniques, such as RNA interference, to target single nucleotide polymorphisms (SNPs) in the ataxin-3 mRNA in linkage disequilibrium with the disease-causing expansion. One of the ways we validated our technology was through its delivery via minimally invasive routes, including intra-cerebrospinal and intravenous administration, using adeno-associated viral vectors (AAVs).

Problem to tackle:

- MJD Disease, or SCA3, is a rare and incurable disease and the most common form of Spinocerebellar Ataxias, affecting up to 2 in 100 thousand adults.
- MJD is a dominantly inherited autosomal neurodegenerative disorder caused by a genetic mutation in the coding region of the ataxin-3 gene (MJD1/ATXN3 gene), which encodes ataxin-3 protein.
- This genetic mutation leads to increased repetitions in the CAG segment of the ataxin-3 gene, with the severity of symptoms dependent on the number of extra repetitions.
- MJD symptoms include slowly progressive clumsiness in the arms and legs, a staggering or lurching gait, difficulty with speech and swallowing, impaired eye movements (sometimes accompanied by double vision or bulging eyes), and lower limb spasticity.



Targeted and minimal invasive gene therapy to treat Machado-Joseph Disease

ADVANTAGES OVER ALTERNATIVE TECHNOLOGIES

- Allele-specific silencing of mutated mRNA while preserving normal ataxin-3 expression
- Single-administration through minimally invasive routes using AAV vectors

APPLICATIONS

Treatment of Machado-Joseph Disease/Spinocerebellar Ataxia type 3 (SCA 3)

PATENT SPECIFICATIONS

Patent: PCT/IB2020/050141
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