ATON - Ataxia Telangiectasia Oligonucleotide Therapy

Introduction

Ataxia Telangiectasia (A-T) is a rare genetic disease caused by mutations in the ATM gene, leading to neurological impairments, immunodeficiency, premature aging, and increased cancer and diabetes risk. ATM dysfunction leads to metabolic disease and imbalanced redox control. Glycogen accumulation resulting from ineffective glycolysis contributes to glucose metabolism imbalances. Accumulation of glycogen was observed also in human neurons and brains, where it might contribute to A-T neuropathology.

Medical Need

A-T is the second most common childhood neurodegenerative disorder of the cerebellum. Premature death is often due to chronic respiratory insufficiency. **No therapies are currently available for A-T**, treatments are only symptomatic and supportive and gene therapy for ATM gene is not expected to work. Patients have a significant unmet need for therapies that can alleviate neurodegeneration and systemic metabolic issues.

Solution

We have **identified a new target**, alternative to ATM gene, that is upregulated in A-T cells, causing metabolic dysfunction and toxicity. Our proposed solution relies on the use of **antisense oligonucleotides (ASOs)** to downregulate the specific target, **restoring normal metabolism** and **enhancing cell viability in A-T cells and neurons**, through a synthetic viability strategy, and blocking neurodegeneration.

Competitive Advantages

- Selectivity
- Specificity
- Effectiveness
- Faster Development (ASO/siRNA: well-establish chemistry and delivery; Orphan Drug Designation should allow a faster FDA approval)

Opportunity

We are seeking **industrial partners and/or investors** to support future development stages of our product, and **out-license** the asset.

Scientific leadership/Inventor

Vincenzo Costanzo, MD, PhD
Director of the DNA Metabolism research program at IFOM, IT
EMBO member since 2006, leader expert in DNA damage response and DNA repair

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