Trial REadiness in Ataxia Telangiectasia (TREAT-AT)

Research Project information

Principal researchers: Dr Rita Horvath and Dr Anke Hensiek
Institute: University of Cambridge, UK
Cost: £250,000 over 36 months co-funding in partnership with Action for A-T (UK), AEFAT (Spain) and BrAshA-T (Australia)
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What are the researchers proposing to do?

There are several challenges to assess the effectiveness of drugs in clinical trials aiming to treat the neurological manifestations in AT:

- Individuals with AT have unique features which can differ between people and may not progress predictably
- There is currently no established test in AT, such as a blood test or brain scan to track changes in the neurological symptoms
- Human samples are needed to better evaluate the effectiveness of an intervention, as the animal models do not present neurodegeneration

The team at Cambridge have identified a particular mutation (the 'UK mutation') that causes a type of AT and may be treatable with a new genetic treatment called antisense oligonucleotide (ASO) therapy. Their study will use 10 years of existing data tracking people with AT and gather a further 2 years of new data in people with the UK mutation.

Why?
This robust and extensive study into the progression of neurological symptoms in AT aims to establish optimal biomarkers in preparation for clinical trials in AT. Biomarkers are biological molecules found in blood, other body fluids, or tissues that are indicators of the disease condition. They can be measured easily and can be used to monitor the disease. Specifically, the team are preparing for a future trial of an antisense oligonucleotide therapy in patients with the UK mutation.

How will the research be done?

The team will study the clinical symptoms, blood biomarkers and brain imaging findings in a subgroup of AT caused by the so called "UK mutation". Team members are experts in all aspects of this study including translational research, state-of-the-art imaging and eye assessment. They will involve AT patients and their family members in the research and also in the design of a future clinical trial. They will also actively engage with AT charities to enable a better distribution of information to patients.

How could it make a difference to the lives of those affected by AT?

This study will help us to understand the natural history of AT and aims to allow rapid progression to trial-readiness, vital to the delivery of clinical trials, such as a novel ASO therapy to a cohort of AT patients. They will ensure the most thorough methods to fairly assess therapy effectiveness, which will benefit any future clinical trial in AT. They will involve AT patients and their family members in the design of a future clinical trial.