

## Natural History of Ataxia Telangiectasia (N-HAT)



### Research Project information

**Principal researchers:** Dr William Whitehouse & Dr Emily Petley

**Institute:** University of Nottingham

**Cost:** £177,865.96 over 24 months in partnership with the A-T Society and BrAshA-T

**Project Completion Date:** 30<sup>th</sup> of August 2022

### Project Overview

Dr William Whitehouse, along with Dr Emily Petley and professionals from the two UK national A-T clinics, aimed to document the natural history of A-T. The NHS set-up in the UK provides a unique opportunity to do such a study.

### Research Methods

Dr Petley reviewed the clinical notes of everyone with A-T seen in the adult and paediatric A-T clinics since 2001, extracting data on infections; immune deficiencies; cancers; lung disease; brain and nervous system disorders; growth; nutritional status; mortality; and genetics. The data was statistically analysed to determine links between signs and symptoms and describe how A-T progresses.

### Project Outcome

The project has documented the pattern of disease of ataxia-telangiectasia (A-T) in 173 people. The outcome has determined at what age manifestations of the disease occur in the UK A-T population, and how common each manifestation of the disease is. This will help doctors and healthcare professionals pre-empt complications of the condition, so that they can aim to treat them early. It is hoped that this will result in a better outcome for people with A-T, for example detecting cancer earlier. This study will also find links between manifestations of the condition.

This study has raised questions and theories that clinicians are currently unable to answer without further research. However, raising these questions and theories will enable further research to be carried out, potentially finding treatments or in the long term a cure for the condition.

By understanding the condition in more detail, the team behind the study hope to help people with A-T and their families to have a greater understanding of what to expect at what age. This will help them to plan their future, for example education, housing, and careers.

Most of the aims of this project were met successfully. COVID-19 did have an impact on meeting with families and the development of focus groups. A plan to approach this in 2023 has been made.

## **Publications**

- Petley E, Yule A, Alexander S, Ojha S, Whitehouse WP. *The natural history of ataxia-telangiectasia (A-T): A systematic review*. PLoS One. 2022 Mar 15;17(3):e0264177. doi: 10.1371/journal.pone.0264177. PMID: 35290391; PMCID: PMC9049793.
- Currently preparing manuscript of descriptive data on natural history of ataxia-telangiectasia.

## **What next?**

Focus groups with people with A-T and their families will be held in 2023 to discuss the results and consult people with A-T and their families about their priorities in A-T research.