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ATG. Ataxia Teleangiectasia

NIHR BioResource – Rare Diseases study project

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Summary

Ataxia teleangiectasia (A-T) is a rare inherited neurological disorder caused by mutations in the *ataxia telangiectasia mutated (ATM)* gene leading to impaired movements of the eyes, limbs and progressive neurological decline in children and adults. *ATM* plays an important role in repairing damaged DNA and the neurological presentations in A-T are often associated with various types of cancer and immunological deficiencies.

Establishing an A-T cohort within the NIHR BioResource will facilitate research on understanding the particular vulnerability of certain neurological systems for altered DNA repair and will explore common molecular pathways across neurodegeneration, immunity and cancer. We will also identify clinical biomarkers that could facilitate clinical trials and the development of treatments in this currently devastating and incurable disease.

Furthermore, *ATM* is also an important tumour predisposition gene, therefore our research can provide insights into cancer research.

Recruitment Criteria

Inclusion

Patients with genetically confirmed ataxia telangiectasia – both the classic and variant form.

Patients with ataxia telangiectasia like disorder (who have a confirmed MRE11 mutation).

Exclusion

None.