



**Application number/Title:** 28541 - Macular Telangiectasia (Mactel type 2) - Systemic, Genetic and Ocular associations

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**Keywords provided by the Applicant PI to describe the research project:**

Eye, Retina, OCT, MacTel, Genetics, Algorithm

**Application Lay Summary:**

1a: Macular telangiectasia (MacTel) is an eye disorder that results in slowly progressive loss of central vision. At present, there is no known cure or treatment to prevent loss of vision in this condition.

Although relatively rare, the true prevalence is unknown. People with MacTel have non-ocular health problems.

We will analyse optical coherence tomography (OCT) retinal images and colour fundus photographs in the UK Biobank database. We have identified genetic changes strongly associated with MacTel. We would like to know how frequently these changes occur in a population and whether they are always associated with eye disease.

1b: The proposed research will utilise existing datasets developed in the UK and with international collaborators. The international MacTel study has a deeply phenotyped group of participants (natural history study, demographics, estimated prevalence based on retinal photograph surveys, genetic studies, family studies, advanced retinal imaging and histopathology, metabolomic, treatment trials). People with MacTel commonly have type 2 diabetes, high BMI and develop eye complications like those in age-related macular degeneration (AMD). We now know that people with MacTel may have a systemic disorder as well. More information about MacTel will improve our knowledge of other human diseases, like diabetes/AMD.

1c: 1. Data cleaning to determine images are assessable by a computer algorithm

2. Assessment of images using machine learning and deep learning algorithms for features of MacTel.

a) Primary outcome – Presence of features consistent with MacTel - calculate disease prevalence

b) Secondary outcomes – Spectrum of disease features on OCT imaging

3. Validation with manual assessment

4. Multivariate statistical modelling techniques to correlate features of OCT

a) Demographic

b) Ocular data

c) Systemic factors and imaging

d) Genetic data

5. Search BioBank genetic database for mutations associated with MacTel on GWAS. Review data for early MacTel or new, non-ocular manifestation/risk.

1d: Full cohort for eye and genetic data. Selected subsets for control groups or other exploratory analyses.

Planned control groups:

1. Type 2 diabetes mellitus - rationale: People with MacTel have a high prevalence of type 2 diabetes.
2. Age-related macular degeneration (AMD) - rationale: people with MacTel develop pathological neovascularisation, a feature of late stage/wet AMD.
3. Exploratory analyses (other may develop during the study) - high BMI (people with MacTel have a higher BMI than matched control groups), evidence of non-ocular neurologic disease (people with MacTel have a retinal neurodegeneration and genetic/metabolomic data suggest possible neural metabolic stressors).