NIHR | BioResource

ION – Inherited Optic Neuropathies

NIHR BioResource – Rare Diseases study project

Lead Investigator: Dr Patrick Yu Wai Man

V1.1 20/05/2019

Dr Patrick Yu Wai Man, ION project Lead

Summary

Inherited optic neuropathies have an estimated prevalence of 1 in 10,000 in the population and they are major cause of chronic visual morbidity in children and young adults. Patients can develop either isolated optic atrophy or a more complicated phenotype with additional neurological features such as hearing loss, myopathy and ataxia.

Inherited optic neuropathies are genetically heterogeneous and they can be caused by both mutations in the mitochondrial genome (e.g. Leber hereditary optic neuropathy, LHON) and the nuclear genome (e.g. autosomal dominant optic atrophy, DOA, and Wolfram syndrome). Despite greater access to genetic testing, about 40% of patients with a clinical diagnosis of an inherited optic neuropathy do not have a confirmed molecular diagnosis, which complicates genetic counselling. Treatment options also remain mostly supportive and there is an urgent need to develop targeted therapies to prevent progressive optic nerve degeneration and visual loss.

The overarching aim of this project is to gain a better understanding of the genotype and phenotype in patients with inherited optic neuropathies, which will help guide optimal clinical design for future clinical trials. To achieve these objectives, we will collect prospective deep phenotyping data on the natural history of this group of disorders and collect blood samples for biomarker profiling or to identify the underlying genetic basis for the patient's optic atrophy.

Recruitment Criteria

Inclusion

- Patient with a clinical diagnosis of a suspected inherited optic neuropathy
- Family member (≥16 years old) of a patient with a clinical diagnosis of a suspected inherited optic neuropathy