NIHR | BioResource

IND – Rare Inherited Neurological Diseases

NIHR BioResource – Rare Diseases study project

Lead Investigator: Prof. Patrick Chinnery

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Summary

Rare Inherited neurodegenerative disorders affect ~1 in 2000 of the UK population. These diseases often cause progressive disability and premature death. There is currently no cure for many of these disorders, placing greater emphasis on genetic counselling to help prevent disease, and further research into the pathogenesis, hopefully leading to the development of new treatment. In many patients it is not possible to make a precise diagnosis at the genetic level. This is extremely important because it allows clinicians to give accurate genetic counselling to patients and their families, and opens up opportunities to develop new treatments based on an understanding of the disease mechanism.

Despite major advances in our understanding of the genetic basis of neurodegenerative diseases, the diagnosis of many familial neurodegenerative disorders remains uncharacterised at the genetic level. Patients attend neurogenetic clinics with a range of inherited neurodegenerative disorders including mitochondrial disease, inherited ataxia, hereditary spastic paraparesis, inherited neuropathy, neuro-ophthalmological disorders involving the visual system, and disorders associated with deafness. Unfortunately, it has only been possible to reach a genetic diagnosis in approximately one third of the patients attending such clinics.

The over-arching aim of this proposal is to identify the genetic basis of these disorders and describe the extent of the clinical phenotype in detail. For the individual patients and their families, this research will:

(i) provide patients with an explanation for their neurodegenerative disorder,
(ii) enable reliable genetic counselling and prenatal diagnosis,
(iii) identify patients with a treatable disorder that cannot currently be diagnosed using standard NHS tests, and
(iv) identify clinical biomarkers that could be used to monitor disease progression.

Based on previous work, it is likely that this will reveal novel disease mechanisms that will potentially lead to the development of new treatments.

Recruitment Criteria

Inclusion

- Clinical diagnosis of a suspected inherited neurodegenerative condition
- Family member, aged 16 or over, of a patient with clinical diagnosis of a suspected neurodegenerative condition
- Children with symptoms of an inherited neurodegenerative condition

Exclusion

Children under 16 with no symptoms of an inherited neurodegenerative condition