Recruiting Research Studies

Inherited Retinal Diseases research at Moorfields

Moorfields Eye Hospital wants to improve access to clinical research studies for all patients within the NHS and provide the opportunities for patients to participate in research.

This is a list of research studies that are currently open and recruiting patients at Moorfields Eye Hospital. This includes information about the study, the lead consultant on the study, and the currently anticipated date that patient recruitment will end. If no end date is stated, then the study has no set end date and will continue to recruit patients for the foreseeable future.

If you are interested in any of the listed studies, we advise that you approach your Moorfields consultant for more information. If you are not a Moorfields patient, you will need to be referred to Moorfields Eye Hospital by your NHS GP to be able to participate in any of these studies. For further information, please e-mail moorfields.resadmin@nhs.net

If a member of our staff thinks you might meet the specific criteria for a study, they will discuss it with you in detail. You might be approached during your visit to the hospital, or receive a letter or phone call after your visit. You will be given time to consider whether you would like to take part and what the implications will be for you. All clinical research projects are strictly monitored by our research department and regulated by national bodies. You will need to give explicit written consent if you decide you would like to take part.
X-linked retinitis pigmentosa (XLRP) is the name given to a group of inherited eye diseases that affect the retina (the light-sensitive part of the eye). Changes in the RPGR genes account for most cases of X-linked retinitis pigmentosa which is more common in men. RP causes the breakdown of photoreceptor cells (cells in the retina that detect light). Photoreceptor cells capture and process light helping us to see and as these cells breakdown and die, patients experience progressive vision loss. The first sign of retinitis pigmentosa is usually night blindness which becomes apparent in childhood or early adulthood and is progressive throughout the subject’s life-time.

There is currently no treatment for RP. Gene therapy is considered the most promising treatment among a variety of novel therapeutic strategies that are currently under investigation. In patients with RP associated with mutations in the RPGR gene, a functional gene replacement may facilitate the functional and morphological rescue of the photoreceptor cells and consequently improved vision. We will include affected people of various ages in this study and children will be included once safety has been established in adults.

Consultant: Prof Jim Bainbridge  
Recruitment End Date: 03/04/2020

BAIJ1016
Long-term follow-up study of participants following an open label, multi-centre, Phase I/II dose escalation trial of a recombinant adeno-associated virus vector (AAV2/8-hCARp.hCNGB3) for gene therapy of adults and children with achromatopsia owing to defects in CNGB3

Study Description (IRAS Form?) The follow up study is designed to collect data on longer-term safety and efficacy at 9, 12, 24, 36, 48 and 60 month time-points following AAV2/8-hCARp.hCNGB3 administration in the CNGB3 trial. The Achromatopsia CNGB3 trial is an on-going open-label, Phase I/II dose-escalation study to determine the safety and efficacy of a subretinal administration of the ATIMP in participants with achromatopsia. In the dose escalation phase, up to 18 adult participants are administered one of 3 different doses of vector in cohorts of 3 participants at a time, using a 3+3 design. Based on toxicity data, the IDMC makes a recommendation on the dose to administer to the next cohort of 3 participants. The IDMC may recommend additional participant are administered any dose before making another decision. Up to 9 children or adults will be included once an acceptable safety profile has been established in adults. The IDMC will agree the maximum tolerated dose in adults before recommending administration of this dose to children.

Safety and efficacy is being assessed by clinical examination and special investigations according to a pre-defined schedule of follow up visits, for 6 months following the intervention. At the 3 month follow up visit of the CNGB3 trial (3 months post-administration), participants will be invited to join this longer-term follow-up study, in which they will be assessed for safety for up to 60 months following AAV2/8-hCARp.hCNGB3 administration.

Consultant: Prof Jim Bainbridge  
Recruitment End Date: 02/03/2020
MICM1002
An Exploratory Clinical and Molecular Genetic Study of Stationary and Progressive Disorders of Cone Function

This research study aims to investigate these conditions in detail to help improve our understanding of the causes of visual impairment in these conditions; to investigate to what degree change occurs over time and to identify the genetic causes of these disorders. This information will assist in the provision of better information for patients and may also be helpful in the event of future anticipated treatment trials. The research aims to improve our understanding of these disorders in order to provide better information to patients and also to shed light on the causes of visual loss in these conditions. We want to establish the genetic cause of these disorders.

Consultant: Prof Michel Michaelides  Recruitment End Date: 31/12/2020

MICM1003
Phenotyping Patients with Leber Congenital Amaurosis Associated with Mutations in AIPL1 in Preparation for Gene Therapy Trials

This research study aims to provide detailed information about retinal function and how this changes over time in patients with AIPL1 gene defects. We hope that this will lead to a better understanding of the condition and assist in providing improved information to patients and also assist in developing future treatment strategies. We want to improve our understanding of the retinal function and structure, and how this changes over time, in patients with AIPL1 gene defects and to be able to provide improved information to patients and also assist in developing future treatment strategies.

Consultant: Prof Michel Michaelides  Recruitment End Date: 31/12/2020

MICM1004
A Study of Conditions Affecting the Retina

The retina is the light sensitive layer at the back of the eye. The eye has many similarities to a camera. The retina is similar to the film in a camera. There are many problems affecting the retina that mainly affect the light sensitive cells called rods and cones, especially in the early stages. Rod cells are mainly used for night-time vision and cone cells are mainly used for day-time vision and reading. We are trying to learn more about the causes of these conditions and look at how well the rod and cone cells work and whether this changes in different people as they get older.

Consultant: Prof Michel Michaelides  Recruitment End Date: 31/12/2020
MICM1006
Magnetic Resonance Imaging Studies for Children with Achromatopsia

The study is being carried out to investigate the relationship between visual cortex structure and function using structural and functional MRI, correlate phenotypic findings with genotype and explore the use of determining efficacy of planned gene replacement therapy using structural and functional MRI and probe the potential role that variable cerebral plasticity may have on limiting treatment effects. The preliminary investigations will aim to determine whether the previously reported cerebral reorganisation that can occur in achromatopsia is age- or genotype-dependent.

Consultant: Prof Michel Michaelides  Recruitment End Date: 31/12/2020

MICM1017
RPE65 Natural History Study

Natural History of patients with Leber Congenital Amaurosis associated with mutations in RPE65

Consultant: Prof Michel Michaelides  Recruitment End Date: 01/02/2021
MICM1021
A Phase 3 Multicenter, Randomized, Double-Masked Study Comparing the Efficacy and Safety of Emixustat Hydrochloride with Placebo for the Treatment of Macular Atrophy Secondary to Stargardt Disease

This study is a multicenter, randomized, double-masked, placebo-controlled study to determine if emixustat reduces the rate of progression of macular atrophy (MA) compared to placebo in subjects with STGD. Subjects will be randomly assigned to one of two treatment arms in a 2:1 (emixustat:placebo) ratio. Subjects will self-administer orally the study drug once a day every evening for 24 months. This study will be conducted at approximately 30 sites in the US, Europe, and other countries. Approximately 162 subjects will be enrolled (108 emixustat and 54 placebo). Randomization and double-masking will be used to minimize bias in subject selection and the evaluation of subjects during the study. A placebo control is included to provide an objective comparison for the safety and efficacy of emixustat. The maximum duration of participation in the study will be 27 months, with a total of 13 visits.

Consultant: Prof Michel Michaelides  Recruitment End Date: 31/05/2020

----

MICM1027
Natural History Study of Patients with X-linked Retinal Dystrophy Associated with Mutations in Retinitis Pigmentosa GTPase Regulator (RPGR)

The purpose of this study is to undertake a detailed prospective phenotypic study of the natural history of retinal dystrophy caused by mutations in the retinitis pigmentosa GTPase regulator (RPGR) gene. Mutations in the RPGR gene are a major cause of X-linked retinitis pigmentosa (XLRP), which is a severe form of retinitis pigmentosa with an early onset of night blindness (generally < 10 years of age) and progression to legal blindness by the 3rd or 4th decade. Mutations in RPGR are also the most common cause of X-linked cone-rod dystrophy (XLCORD), where affected males develop onset of visual symptoms from the 2nd to 4th decade, and show progressive declines in central vision followed by declines in night vision, development of light sensitivity, and myopia. RPGR mutations have also been shown to be associated with X-linked recessive atrophic macular degeneration. Proof of principle studies for RPGR gene replacement therapy has been demonstrated with gene replacement in an RPGR mouse model.

Human clinical trials are now underway. We intend to undertake a detailed prospective phenotypic study to investigate the natural history of RPGR. Detailed phenotyping of patients with RPGR mutations should facilitate the identification of an optimal window for intervention, provide specific parameters to quantify treatment effects and define clinical endpoints, and will help identify suitable patients for therapeutic intervention.

Consultant: Prof Michel Michaelides  Recruitment End Date: 01/04/2022
Invitro modelling of early onset retinal degeneration by derivation and differentiation of human induced pluripotent stem cells

In this project we propose to model early onset or congenital retinal dystrophy in the laboratory by generating photoreceptor cells from a tissue sample derived from affected individuals using a new technology involving induced pluripotent stem cells (iPSC). We will harvest human fibroblasts, peripheral blood cells or exfoliated renal epithelial cells present in urine from patients with congenital or early onset retinal degeneration and known gene defects. We will reprogramme these cells using iPSC technology and derive photoreceptor cells via the formation of ocular cups, culture these in the laboratory and characterise the effect of the gene defects on their phenotype by comparison with cells from normal control individuals. The primary purpose is to test the feasibility of this approach to model early onset or congenital retinal dystrophy and to investigate the causative relationship between specific gene defects and the disease phenotype. We anticipate that a better understanding of how gene defects lead to retinal degeneration may lead to the identification and development of therapeutic interventions to preserve vision and prevent blindness.

Consultant: Dr Jaqueline Van der Spuy  
Recruitment End Date: 01/03/2022

EURO-WABB: An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome, Bardet-Biedl Syndrome and Other Rare Diabetes Syndromes

Wolfram, Alström and Bardet Biedl (BBS) (WABB) syndromes are rare genetic diseases with clinical overlap, chronically debilitating, and highly complex. Patients are distributed throughout the EU. The diseases progress to death in early adulthood. EUROWABB is an international registry of children and adults WABB and other rarer diabetes syndromes, containing anonymised clinical data on history and examination, complications, laboratory investigations, imaging; and molecular genetic data; and information on human tissue held locally. The purpose of the web-based registry is: a) to establish the natural history of these diseases (their characteristics, management and outcomes); b) to assess clinical effectiveness of management and quality of care; c) to provide a register of patients for recruitment to intervention studies; d) to establish genotype-phenotype correlations. Data held in the registry will be anonymised (there will be no personal identifiable data held); but linked by a unique identifier to the health professional who submitted the data. The local health professional can submit data records on his/her patients, and controls access to data records for patients under their care. Patients will be able to submit some of their own personal data if they want. The detailed clinical characterisation will allow future enrolment into multinational clinical trials (with separate consent). The registry will support wider access to genetic testing, information for affected families and health professionals; and encourage international collaborations for patient benefit.

Consultant:  Mr Patrick Yu Wai Man  
Recruitment End Date: 31/12/2021