3 Year PhD Studentship available for September 2019

**Department:** Neuroscience, Psychology and Behaviour

**Supervisors:**
First supervisor: Dr Mervyn G Thomas  mt350@leicester.ac.uk
Second supervisors: Professor Irene Gottlob ig15@le.ac.uk and Professor Eugene Yu-Dong Zhang  Yudong.zhang@leicester.ac.uk

**Eligibility:** This studentship is open to UK and EU applicants only.

**Project Title:** Identification of novel genes and addressing the missing heritability in albinism

**Project Description:**

Albinism is a genetically heterogeneous group of congenital hypomelanotic disorders. It is characterized by ocular features such as infantile nystagmus, iris transillumination, foveal hypoplasia, chorioretinal hypopigmentation, photophobia and axonal misrouting at the optic chiasm. The cutaneous manifestations are characterized by skin and hair hypopigmentation. Syndromic forms of the albinism, although rare, can be life threatening. These include Hermansky Pudlak syndrome and Chédiak-Higashi syndrome. To date, 19 genes have been described to be associated with albinism. However, these genes only account for approximately 70% of mutations associated with albinism. Thus, a significant proportion (30%) of cases remain unsolved. We hypothesize these cases are likely due to novel disease genes and potential disease modifying variants associated with albinism that are yet to be discovered. We propose the use of next generation sequencing (NGS) and machine learning based bioinformatics techniques in these unsolved cases for novel gene discovery and variant characterisation. This project will be closely linked to the 100,000 genomes project where we will be spearheading the analysis of genomes in patients with infantile nystagmus and albinism.

The ophthalmology group at Leicester has extensive experience in genotype-phenotype studies in infantile nystagmus and has access to one of the largest cohort of patients with infantile nystagmus. We have developed the first NGS panel for nystagmus which tests for over 336 known genes associated with nystagmus including all known albinism genes. The successful candidate will utilise our gene panels and whole genome/exome sequencing to genetically characterise a cohort of patients with albinism. We will also have access to data from the 100,000 genomes project for novel gene discovery and variant analysis. The
candidate will be trained in use of the high performance computing cluster (ALICE) and specific pipelines which utilise machine learning to analyse and interpret NGS data.

**Key methods in which the student will receive training:**
DNA extraction  
PCR  
Sequencing  
Bioinformatic analysis – use of high performance computing cluster (ALICE), machine learning protocols

**References:**


**Funding details:**

This project is in competition for a College of Life Sciences (CLS) PhD Studentship. The Studentship is for three years, starting September 2019, and offers tuition fees at UK/EU rates and a Stipend at UK Research Council rates.

**Entry requirements:**

Applicants are required to hold/or expect to obtain a UK Bachelor Degree 2:1 or better in a relevant subject. The University of Leicester English language requirements apply where applicable.

**How to apply:**

You should submit your application using our online application system. We are unable to consider applications submitted by email.  

Guidance for completing the online application form: Under ‘Select your area of study,’ choose ‘Neuroscience Psychology and Behaviour Research’; under ‘Select your intake date,’ choose ‘September 2019’.  

In the Funding section of the application, select please state you wish to be considered for a NPB College of Life Sciences Funded Studentship.

In the Research Proposal section of the application, please provide the name of the supervisor and project you want to be considered for. You do not need a research proposal
but you should submit a personal statement explaining why you are interested in this project.

**Project / Funding Enquiries:** Applicants are encouraged to contact prospective supervisors by email or phone to discuss the project and their interests prior to submitting a formal application.

Application enquiries to [pgradmissions@le.ac.uk](mailto:pgradmissions@le.ac.uk)

**Closing date for applications:** Monday 21 January 2019