WTDTP Projects for September 2022

Project Reference: T3/41

Project Title: **Unravelling the genetic basis of normal and abnormal retinal development**

Theme(s): Theme 3: Genetically Informed Causal Inference and Risk Prediction

Primary Supervisor: Dr Mervyn Thomas (University of Leicester)

Secondary Supervisor(s): Prof Martin Tobin (University of Leicester)

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Project Summary: Humans achieve high spatial visual acuity due to the fovea. Developmental arrest of this area, foveal hypoplasia, often this arises due to variants in genes expressed early in retina. This has lifelong consequences of poor vision, nystagmus (oscillating eyes) and negative social stigma – often with most individuals having vision below driving standards. Optical coherence tomography allows non-invasive high-resolution foveal visualisation and is routinely used in clinical care. Leicester was the first centre in Europe to pioneer its use in paediatrics. We have developed grading systems and artificial intelligence-based techniques to classify the degree of arrested retinal development. The Leicester grading system for foveal hypoplasia has both diagnostic and prognostic implications and used routinely as part of clinical care. However there remains significant knowledge gaps in our understanding of the genetics of foveal hypoplasia. The team in Leicester leads the largest consortium on foveal developmental studies “Foveal Development Investigators Group (“FDIG”) consisting of 11 centres across 9 countries aimed at improving our understanding on the genotypic and phenotypic correlations in foveal hypoplasia. The successful candidate will work on unravelling the genetic basis of foveal hypoplasia using sophisticated bioinformatic and machine learning techniques using large datasets from both public and local/collaborative datasets.