Application number/Title: 21677 - Genetic and environment studies of myopia and refractive error in the UK Biobank

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Keywords provided by the Applicant PI to describe the research project:

Epidemiology, genetics, myopia, refractive-errors, vision

Application Lay Summary:

Refractive errors, which include myopia (short sightedness) and hyperopia (farsightedness), are the most common causes of visual impairment worldwide. Myopia has become much more common worldwide in recent decades; in 2010, about 1.45 out of 6.5 billion people had myopia. The ?myopia epidemic? has been attributed to rapidly-changing modern lifestyles in many parts of the planet. Nevertheless, genetics explains more than 60% of the range in refractive errors in the population. We will identify both genes and environmental factors to learn more about myopia and stimulate future improvements in its control and management. This research will adhere to the UK Biobank's primary mission of improving the prevention, diagnosis and treatment of serious illnesses - including eye disorders. We will study the most important cause of functional vision impairment in the UK and worldwide (refractive errors, including myopia and hyperopia) and which is an important global public health problem. Myopia has increased rapidly in incidence within the last 3 decades, and population research has linked it to dozens of environmental and genetic risk factors. This research offers a unique opportunity to comprehensively study the effect of genes and environment on refractive error. Refractive errors (RE) measured with an automated device will be available for ~130,000 study participants. In addition, the age at which participants first wore glasses or contact lenses will be available for ~460,000 individuals. We will identify socio-demographic, environmental, and biological risk factors associated with the severity of RE and myopia age-at-onset. Through established genome-wide association study methods, we will also identify the genes responsible for optical development of the eye, including rare mutations which can cause severe, blinding, myopia and hyperopia. We are requesting the full cohort genetic data: data from participants with either automated refraction data (n?130,000) or questionnaire data relevant to

refractive error (n?470,000) will be used in our analyses. We will also request genetic data from all participants to conduct genome wide association studies, estimate rare variant allele frequencies, and identify individuals with potential rare loss-of-function variants within refractive error loci. No biological samples will be requested for this phase of the study.