UK Biobank Eye & Vision Consortium

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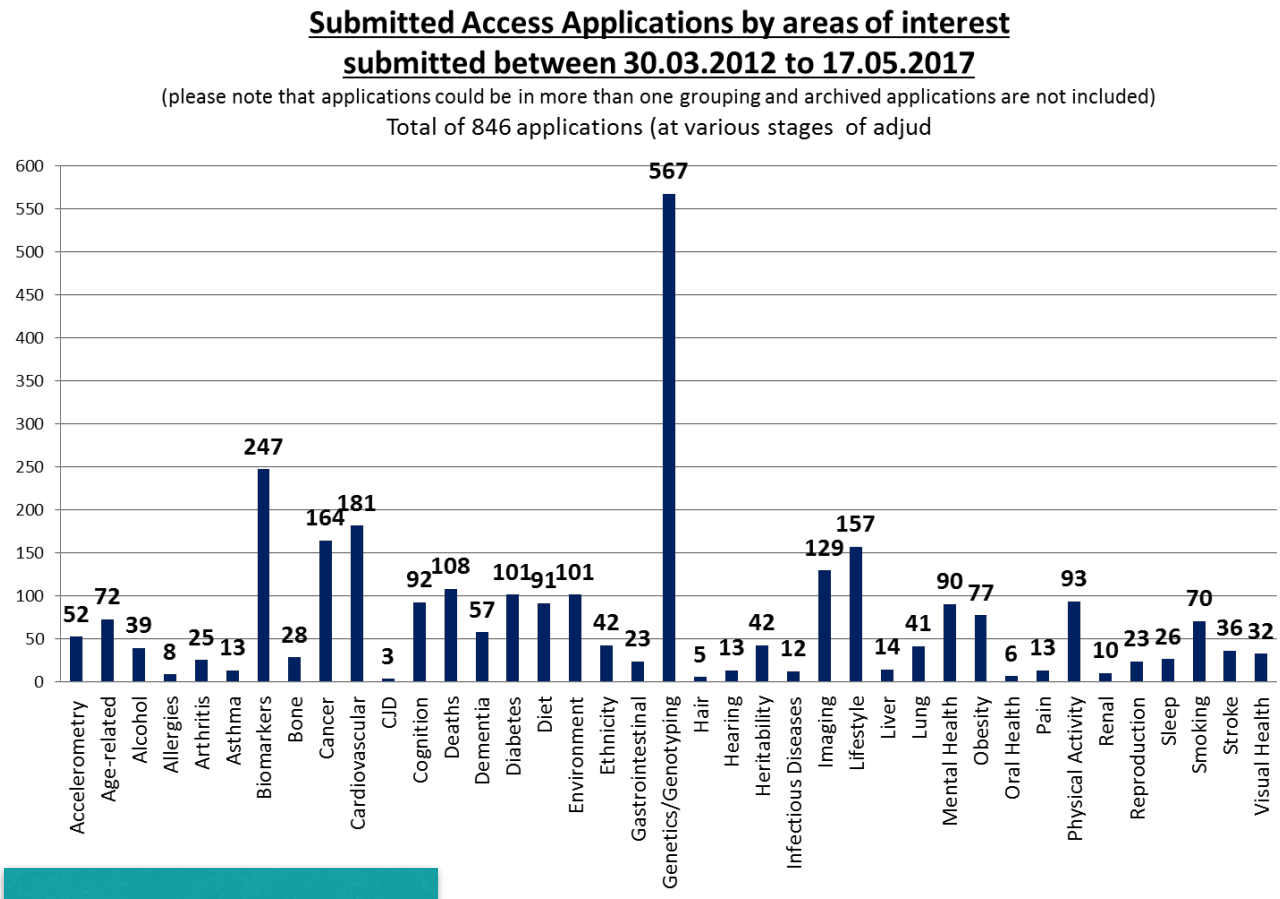
Release of genetic data



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The main event for UK Biobank this year has been the release of the full genetic dataset (20th July 2017). This has excited considerable interest, with applications for a genetics-focussed analysis now the run-away leader in terms of data access applications.

The genetics data has led to a surge in interest in eye-related data from outside the UK. In addition to the data application from Google that was discussed at our 2017 meeting, a group led by Dr Xiaoyi Gao at The University of Illinois at Chicago had their access request approved by UKBB on 4th August 2017, aiming to "identify

novel single nucleotide polymorphisms (SNPs) and pathways associated with ocular phenotypes such as glaucoma, cataract, macular degeneration, intraocular pressure, and corneal hysteresis". A group from China led by Prof Zi-Bing Lin in Wenzhou have applied to run a GWAS of ocular disease in the UK and East Asia (access request granted January 2017). Bayer have requested access UKBB data to "identify and validate new drug targets for diseases … of the eye, the cardiovascular, respiratory and reproductive organ systems, as well as various types of cancers, e.g. in Immunooncology," as well as in autoimmune, immunological and chronic inflammatory conditions. Also John Witte at UCSF applied for a project extension to widen the scope of a study of cancer genetics to include glaucoma and macular degeneration. These two eye conditions also feature in a data request from Goncalo Abecasis at University of Michigan. A joint Stanford-Finlandic group ran a hypothesis free analysis of genetics and hospital linkage data (ICD-10 codes) with the declared aim of identifying novel "drugable" targets, with a particular interest in protein truncating variants (PTV's).

The story gets interesting when one reaches the data request (approved 5th July 2017) from the QIMR in Brisbane, and long-time eye genetics researcher Stuart MacGregor. This application declares an interest in using GWAS data "already in hand" and UK Biobank data to examine "the genetics of various cancers, eye disease and psychiatric traits". Communication between UKBB Eye and Vision Consortium members has revealed that a group in Australia was aiming to publish on genetics of glaucoma related traits imminently. Pirro Hysi and Anthony Khawaja have been working on a similar analysis in collaboration with the NEIGHBORHOOD Consortium, led by Janey Wiggs (Cambridge, Ma), and Nick Wareham's group (MRC Epidemiology Unit, Cambridge, UK).

Pirro and Anthony have submitted a strong manuscript which appears to increase the number SNP's implicated in IOP significantly. We wish them the best of luck with this submission.

Next UKBB Eye & Vision Consortium Annual Meeting:

TUESDAY 6TH FEBRUARY 2018

Wellcome Collection
183 Euston Road, London NW1 2BE

– Agenda items –
Imaging updates, genetics updates, Claire Tochel (UKBB – Outcomes) to speak.
Any other items to Paul Foster please.

- If you wish to attend, please save this date -

UK Biobank Imaging Study

The imaging study, aiming to perform heart, brain and abdomen MRI's, DEXA scans and carotid ultrasounds on 100,000 UKBB participants was featured in the last newsletter. Representations have been made to UKBB to make the case of including eye imaging in this study. It was encouraging therefore, when Paul Foster and Chris Owen were invited to participate in the Dementia Platforms UK (DPUK)'s Re-imaging Proposal which aims to secure funding for re-scanning of 10,000 of the initial 100,000 people in the imaging cohort. There was strong support for including eye measures in the Re-imaging Study, and this has spurred a re-examination of the feasibility of including eye measures in the main imaging study.

Outcomes Adjudication

Parul Desai has been in discussion with UK Biobank regarding the outcomes adjudication from hospital linkage data. Initial efforts will focus on cataract, AMD, glaucoma and diabetic retinopathy. We request volunteers to form working groups to advise on these four conditions. Anyone who is interested please contact Parul Desai or Paul Foster.

New Members

We would like to welcome these new members to the E&V consortium:

Marcus FRUTTIGER
UCL Institute of Ophthalmology

Euan PATERSON
Queens University Belfast

Sarah ENNIS & Jay SELF
University of Southampton

Sobha SIVAPRASAD
Moorfields Eye Hospital, London

PUBLICATION NEWS

Vitreoretinal interface abnormalities in middle-aged adults with visual impairment in the UK Biobank study: prevalence, impact on visual acuity and associations. McKibbin M, Farragher T, Darren S on behalf of the UK Biobank Eye and Vision Consortium. April 2017. **BMJ Open Ophth** 2017;1: e000057. Link to PDF.

Automated arteriole and venule classification using deep learning for retinal images from the UK Biobank cohort. Welikala RA, Foster PJ, Whincup PH, Rudnicka AR, Owen CG, Strachan DP, Barman SA, on behalf of the UK Biobank Eye and Vision Consortium. Sept 2017. **Computers in Biology and Medicine**, 90 pp. 23-32. Link to PDF.