

## EM 9

Expanding DPUK Genetics and integrating with inflammation/immunity research				
Start date: Apr 2018			Completion date: 30 Apr 2020	
<b>Overall objective:</b> This pilot study will genotype samples from across the DPUK cohorts and use state-of-the-art statistical analytics produced in-house to produce an exemplar statistical model of AD prediction.				
Deliverables	Milestones	Milestone deadline	Work package dependencies	Person(s) responsible
<b>Objective 1:</b>				
D1.1 -Selection of appropriate cohorts	M1.1.1 Appropriate individuals identified with rationale for selection clear	M1.1.1 Complete		Williams, Menzies, Sims
D1.2-Preparation of DNA from selected individuals in the cohort	M1.2.1 Genotyping complete	M1.2.1 Complete		Williams, Menzies, Sims
<b>Objective 2:</b>				
D2.1- Calculation of polygenic risk score	M2.1.1 Completion of work on polygenic risk scores	M2.1.1 Dec 2019	Depended on M1.2 completion.	Williams, Menzies, Sims
D2.2-Statistical model available	M2.2.1 Statistical modelling completed	M2.2.1 Feb 2020	Dependent on D2.1	Williams, Menzies, Sims
D2.3 Statistical summaries produced	M2.3.1 Statistical summaries available	M2.3.1 Mar 2020	Dependent on D2.2	Williams, Menzies, Sims
<b>Updates on delivery against milestones since last report</b>				
<ul style="list-style-type: none"> <li><b>M1.2.1 Genotyping complete (Feb 19) – Sept 19</b></li> </ul> <p>Genotyping was held up due to issues with collaborator MTAs. These have now been resolved.  324 MRC samples genotyped to date.  702 MRC samples still to be genotyped. Samples undergoing QC. Genotyping to take place over the next month.  1392 Janssen (Chariot-Pro) samples to be genotyped.  Total number of samples = 2418  Once all data is available the statistical model of AD prediction can be generated.</p>				
<b>Summary of plan to deliver on outstanding work (with dates)</b>				
<p>All material transfer agreements in place &amp; fully executed.  Genotyping of 324 MRC samples on GSA is complete.  MRC samples still to be genotyped are undergoing QC. Genotyping to begin imminently. Expected completion Dec 2020  Janssen Chariot-Pro samples to be shipped to Edinburgh imminently. Completion of genotyping expected by Dec 2020</p>				
<b>Risks</b>		<b>Mitigation</b>		
1) Janssen Chariot Pro samples not yet at Edinburgh.		1) Requested samples be sent to Edinburgh as soon as possible to avoid further delay.		
<b>Team members funded (full or part-time) by DPUK</b>				
(N/A)				

**Team members involved with the project but not funded by DPUK**

Julie Williams, Beccy Sims, Georgina Menzies

**Outcomes**

Awaiting genotyping data for 2094 samples (MRC and Chariot Pro).

Data from genotyped MRC samples undergoing QC – 324 samples.

Extra 418 samples included in project compared to project application.

**Project narrative**

Following the delay with the MTAs, these were finalised in late summer, meaning the Janssen Chariot-Pro samples could finally be shipped for genotyping.

Since the initiation of the project, new genotyping platforms have become available which has reduced the cost per sample for genotyping. This has allowed a further 418 samples to be genotyped. 324 have already been done. The remainder are undergoing QC and genotyping will commence imminently.

The involvement of industrial partner Cytox was not pursued.