

Neurodegeneration & Genomic Sciences

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At TheGBLab our work is focused on understanding genomic variability and its impact on disease/phenotype. Our main areas of interest are neurodegenerative diseases in general, with a particular focus on the most common diseases: Alzheimer's, Parkinson's and Dementia with Lewy Bodies. To study these conditions we apply state-of-the-art genomic technologies in both large cohorts of cases, and on families where these diseases segregate. The former approach tends to inform us regarding disease modulating variants, while the latter is suited to identify disease causing variants. In addition to disease-based research, we also have a longstanding interest in the role of benign genetic variability.

Alzheimer's disease genetics

The search for genetic risk factors in Alzheimer's disease had another positive year. We co-authored a large-scale [study](#) showing for the first time that low frequency variants in PLCG2 and ABI3 modulate risk for AD. These are important results because they continue to highlight microglia as an important player in AD pathogenesis. In that same study we continue to find evidence for risk at the TREM2 locus in AD. Because of the specific link between TREM2 and TYROBP at the protein level, we tried to determine if genetic variability at that gene is involved in risk for AD. We performed this [analysis](#) using the same cohort that led to the initial discovery of TREM2 in AD, but, in this case, found no evidence of genetic risk at TYROBP.

To identify rare genetic variability in well characterised AD samples, we [published](#) a preliminary analysis of the [Brains for Dementia Research](#) cohort. This work is ongoing and we aim to complete sequencing of the entire cohort in 2018. Also in our cohort studies, together with our colleagues at the Dementia Research Center, we [published](#) the study protocol of the Insight-46 cohort, for which we are leading the genetic analyses.

Lastly, we co-authored a [manuscript](#) showing evidence for a locus conferring genetic resilience to AD. A genetic variant at RAB10 was found to confer significant protection against AD, which could open novel avenues for future therapeutic approaches.

Dementia with Lewy Bodies genetics

In DLB, 2017 will go down as a milestone year for us. The year was bookended by DLB papers with a couple of very important ones in between. The [first](#) showed that C9orf72 expansions are not a common cause of DLB in a large neuropathologically diagnosed cohort. We then went on to co-author the [fourth consensus report of the DLB consortium](#). These have always been landmark papers that guide the diagnosis and

management of DLB patients and have always been expertly led by Ian McKeith. We co-authored a [perspective](#) on the molecular differences and similarities between DLB and PDD taking into account recent data.

And lastly, we [published](#) the first genome-wide association study in DLB. This was the first large-scale genetic study in this disease that showed that common genetic variability at several loci modulates risk for the disease. It also allowed us to start to see differences and similarities in genetic risk profiles between DLB, PD and AD. All in all, a very important paper for us that received great coverage [here](#), [here](#) and [here](#).

Rare diseases

Despite our main projects being in common neurodegenerative diseases, one area which we have always had an interest in is the study of rare or orphan diseases. 2017 was no exception. We saw the [identification](#) of pathogenic mutations in TBC1D24 in a sibship with Multifocal Polymyoclonus. The only described case of Schaff-Yang syndrome in Portugal was also [published](#) this year as part of an international collaboration. We [published](#) a rare, non-coding mutation in COL4A1 as the cause of multi-infarct dementia of the Swedish type. And lastly, we co-authored a [manuscript](#) showing a novel mutation in ZFH2 as the cause of pain insensitivity in a single family. This paper received great coverage [here](#) and [here](#).

Dementia Research Institute

One of the first important events of the year was the news that the [DRI](#) Hub had been awarded to [UCL](#). The DRI is a new, national research institute dedicated to understanding the underlying diseases, transforming treatments and improving the lives of people living with dementia. It has a starting budget of £250 million and is led by the Medical Research Council with founding charity partners (Alzheimer's Society and Alzheimer's Research UK). Shortly after this announcement, Bart de Strooper was appointed director of the DRI. We applied for a Programme grant on the first wave of applications. After a rigorous, very competitive, externally peer-reviewed process we were awarded one of the first DRI Programme Grants to continue our study of genetics in neurodegenerative diseases. This was a real achievement and will allow us to expand our work looking at the effects of genetic variability on brain disorders.

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