



# Functional Gene Annotation Initiative

Using the Gene Ontology to describe proteins and microRNAs

Newsletter March 2021

## Introduction to Bioinformatics and GO workshop – 14<sup>th</sup>-15<sup>th</sup> June 2021

If you would like to attend our popular free two-day workshops, please use the [Eventbrite](#) site to register. This workshop provides an overview of several biological knowledgebases, as well as an introduction to GO and functional analysis tools.

## Welcome to new staff and students

I am very pleased to welcome three new students, a volunteer and a new member of staff to the team. Kate Thurlow is currently funded for two months curating Alzheimer's Disease susceptibility proteins as part of the ARUK funded project; she is currently focusing on curating phospholipase C gamma 2 (PLCG2) research. Her review of over 100 articles supports the findings by [Poux et al., 2017](#), that manual annotation is scalable, as only 6 of these articles provide data suitable for GO curation. Although, during curation it is possible that not all articles will provide novel information not previously associated with PLCG2. Kate undertook an MSc annotation project with the team as part of her MSc in Genetics of Human Disease, for which she received a distinction, so I am pleased to welcome her back. Two MSc students have joined the team, Diana Luna Buitrago and Miao Long, and they are focused on microRNAs that regulate the 'bad' amyloid-beta receptors and tau-associated processes respectively. I am pleased that Marios Makris enjoyed his project so much that he has returned as a volunteer to annotate microRNAs regulating transporters at the blood brain barrier. Additionally, I welcome another BHF PhD student, Yue Jiang, who is joining the team for 3 months and is keen to improve the GO annotation of T2D risk loci associated proteins.

## Annotation progress

Since the last newsletter, Penelope Garmiri (EMBL-EBI curator funded by ARUK) has curated two genes, [ABI3](#) (NESH) and [OAS1](#) by capturing recent information based on 7 and 15 articles, respectively. ABI3 is a member of the SCAR complex and Penelope has captured its role in regulating actin polymerization and depolymerisation and in lamellipodia assembly. Of interest to the Alzheimer's research community is the role of ABI3 postsynaptic density assembly and dendritic spine morphogenesis. In contrast, OAS1 has been linked with surfactant homeostasis in alveolar macrophages and one isoform of OAS1 was found to have a positive impact on mitochondrial respiration in response to interferon-beta. The OAS1 gene cluster region has been associated with severe Covid-19 ([Pairo-Castineira et al. 2020](#)) and the role of the OAS family in antiviral innate immune response maybe relevant this.

Penelope also captured an interesting association between the OAS1 and another AD-associated risk gene, [SPI1](#). The transcription factor, PU.1 (SPI1), regulates the expression of several pro-apoptotic interferon-stimulated genes, including OAS1 and TRAIL ([Goto et al. 2017](#)) and is required for the differentiation of a variety of cell types from the myeloid and lymphoid lineages including microglia, pericytes and pro-T cells.

In total, [all ARUK-UCL](#) projects have resulted in 14,686 annotations for 2,723 distinct gene products, of which 10,990 annotations are associated with 1,900 [human gene products](#) (23 February 2021). Over the past 13 years the UCL functional gene annotation team has submitted over 74,276 annotations to 10,094 distinct gene products, through the review of over 7,500 articles. Despite this volume of annotations, it is difficult to assess the impact of this work on data analysis. However, it is good to see that the UCL team has made a significant contribution to the GO annotation of one of the key proteins recently thrown into the spotlight during the current pandemic ([ACE2](#)), SARS-CoV-2 enters host cells via this receptor ([Lan et al. 2020](#)). This protein has 178 annotations, of which UCL provided [29](#), UniProt [41](#) and IntAct identified [67](#) entities that ACE2 interacts with.

## Goodbye to Chloe

During her rotation project, Kan Yan Chloe Li, provided over 300 annotations for almost 60 proteins, capturing their role in sinoatrial and atrioventricular node development. Chloe has now moved on for her second rotation project with Professor Shanie Mahadeo and I wish her every success in her next project.

## Recent publications

Gene Ontology Consortium. **The Gene Ontology resource: enriching a GOLD mine.** Nucleic Acids Res. 2021 Jan 8;49(D1):D325-D334. PMID: [33290552](#).

Porrás P, et al. **Towards a unified open access dataset of molecular interactions.** Nat Commun. 2020. 11(1):6144. PMID: [33262342](#).

Contact [r.lovering@ucl.ac.uk](mailto:r.lovering@ucl.ac.uk) to subscribe to this newsletter or follow us [@UCLgene](#)  
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