



BiG Talks - Bioinformatics and Genomics Seminar Series

Arranged by the SciLifeLab platforms for Bioinformatics, Genomics and Diagnostics Development

The Bioinformatics and Genomics seminar series (“BiG Talks”) is a new initiative arranged by the SciLifeLab platforms for Bioinformatics (NBIS), Genomics (NGI) and Diagnostics Development. The seminar series aims to give inspiration to the SciLifeLab community and to create new networking possibilities. The BiG Talks will rotate between different SciLifeLab nodes and the aim is to have these seminars four times a year. Also, each event will be live broadcasted at the [SciLifeLab YouTube channel](#) for people outside the different sites to be able to join in.

The first BiG Talk event will take place at SciLifeLab Uppsala (Trippelrummet, Navet) on June 11th and we welcome everyone that are interested in Bioinformatics and Genomics to join in. After the seminar (around 11.30) a lunch sandwich will be served for those who are registered for the event. During the lunch there will be a chance to mingle with the speaker and bioinformaticians and other interested people in the seminar topics. Also, we would like to hear your feedback for the BiG Talk initiative to make this seminar series as successful as possible. Please follow this link to register for the lunch sandwich for the Uppsala seminar 11th of June: <https://goo.gl/forms/e4ZbBHBURH0tWyqW2> (Deadline 6th of June).

Planned Schedule for 2018:

11th of June, 10:30-11:30, SciLifeLab Uppsala

Dr. Paul Lacaze, Head of the Public Health Genomics Program, Monash University, Melbourne, Australia

Title of presentation: Genome sequencing of 15,000 healthy elderly Australians: implications for clinical genetics

1st of October, SciLifeLab Stockholm

Dr. Paolo DiTomasso, Centre for Genomic Regulation (CRG), Barcelona, Spain

Title of presentation: Enabling reproducible in-silico data analysis with Nextflow

Late November, early December: To be announced



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Genome sequencing of 15,000 healthy elderly Australians: implications for clinical genetics

Dr. Paul Lacaze, Head of the Public Health Genomics Program, Monash
University, Melbourne Australia

Abstract:

The ASPREE Healthy Ageing Biobank contains ~15,000 consented samples from individuals aged 70 years or older participating in the ASPirin in Reducing Events in the Elderly (ASPREE) study - Australia's largest clinical trial and longitudinal study of healthy ageing. At enrolment, all ASPREE participants were confirmed to be free of major life-threatening cardiovascular disease, cancer or cognitive decline, meaning samples were ascertained from confirmed healthy elderly individuals, depleted of typical monogenetic disease phenotypes. All ASPREE biobank samples are being sequenced using a targeted 'super-panel' of 750 genes used commonly in clinical testing, including all ACMG59 genes plus pan-cancer, cardiovascular and neurological gene coverage. Over 11,500 samples have been sequenced (May 2018), identifying hundreds of actionable pathogenic variants in individuals lacking any apparent signs and symptoms of genetic disease beyond 70 years. Results will be presented on these findings, with implications for our understanding of penetrance and clinical actionability for genes used in routine testing. In addition, ASPREE has conducted whole genome sequencing on 3000 of the oldest, cancer-free Australian participants as part of the Medical Genome Reference Bank (MGRB) project. This presentation will give an overview of the primary ASPREE trial, depth and breadth of longitudinal phenotype data, and programs of genomic research.

Biography: Paul Lacaze is the inaugural Head of the Public Health Genomics Program at Monash University School of Public Health and Preventive Medicine. He specialises in large-scale genetic analyses of cohort studies, biobanks, clinical trials and clinical registries. He leads the genomic analyses of the ASPREE Biobank and has research interest in the identification of rare cases of non-penetrance or 'resilience' against known pathogenic variants in the healthy elderly. Dr Lacaze also conducts research into the ethical, legal and social implications of genomic information in society. He has a PhD from the University of Edinburgh and experience in the commercial and academic life-science sectors.

Host: Adam Ameer, NGI Uppsala (adam.ameur@igp.uu.se)

Date: June 11, 10.30-11.30 in Trippelrummet, Navet, SciLifeLab Uppsala and online

Broadcast link (live event): [SciLifeLab YouTube channel](#)

Registration for lunch sandwich: <https://goo.gl/forms/e4ZbBHBURH0tWygW2> (Deadline 6th of June).