

Speaker: Dr Sara Ballouz, Garvan Institute of Medical Research



Biography:

Dr Ballouz central scientific interest has been to understand the genetic architecture of disease. With data from the genome, transcriptome, epigenome and proteome increasing exponentially, robust tools and practices need to be established to analyze this deluge, in particular if to be applied to personalized medicine. The group's current focus is on exploring and understanding X-linked disorders and sex differences in disease, with a particular interest in autoimmunity.

Abstract:

Up to 12% of the Australian population suffers from an autoimmune disorder, with approximately 80% of patients being female. Symptoms are often broad (e.g., fatigue, rashes, pain) and can sometimes overlap across diseases and disorders. These co-morbidities firstly make diagnosis difficult for clinicians and secondly frustrating for patients, as it can take decades to obtain the correct diagnosis. Anecdotally, self-diagnosis, self-medication and reliance on community groups to receive support are common. From a research perspective, this introduces confounding factors when investigating genomic signals, as well as the efficacy of clinical trials downstream.

To characterise the frequency of these practices and how they might impact our genomic research efforts, we have launched a survey around chronic illness diagnostic journeys. We aim to explore the respondent's submissions for connections around mental health, quality of life, fatigue scores and symptoms to characterise differences and similarities of diagnostic journeys and medication in chronic illness. Additionally, this survey will help us recruit patients for future genetic and genomic studies.

While still in progress, we have received over 1000 responses across Australia to date, with a combination of over 160 chronic illnesses. The majority of respondents are female (76.6%), which follows with current estimates on prevalence. Further to this, using network analysis tools, we have been able to build a classifier to assess health status and present some of the insights from this preliminary work. The study continues to run with information available at <https://spoonie-community.netlify.app/>. We are also looking for controls, so even if you do not have any of the specified health conditions, we would also be interested in your general responses. If you would like to participate, it should take between 10-20 minutes and can be accessed from the link above.

All welcome!

[Click here to join the meeting](#)

Enquiries: Lindsay Wu lindsay.wu@unsw.edu.au