Centre for Health Policy

PhD Confirmation Seminar

The University of Melbourne

Melbourne School of Population & Global Health

The health economics of functional genomics for undiagnosed rare diseases in Australia

Presenter: Francisco Santos

Supervisors: Associate Professor Ilias Goranitis (Primary Supervisor, University of Melbourne), Co-supervisor Professor John Christodoulou (Murdoch Children's Research Institute)

Friday 10 February 2023, 11am-12:00pm (AEDST)

Seminar Room 405a
Level 4 207 Bouverie Street
Carlton, VIC 3053

Via Zoom:
Meeting ID: 813 7920 1164
Password: 632413

Nearly 80% of rare diseases have a genetic origin. Yet almost half of Australian patients are not diagnosed despite advances in genome sequencing. There is potential for functional genomics, such as transcriptomics and proteomics, to overcome the diagnostic challenges of genome sequencing. However, the implementation of functional genomics diagnostics in the Australian healthcare has not yet been backed by health economic evidence. This PhD aims to address this research gap in four sequential phases, starting with micro-costing both functional genomic platforms to estimate delivery costs, followed by evaluating the economic implications of clinical utility based on clinician surveys. Functional genomics effectiveness will then be evaluated by obtaining diagnostic outcomes as well as personal utility and health-related quality of life changes. These findings will be incorporated into an economic evaluation model to assess the cost-effectiveness and cost-benefit of functional genomic tests in patients with rare diseases.

Francisco is a PhD student in Health Economics at the Melbourne School of Population and Global Health. His PhD research project aims to develop a health economic evaluation framework for the implementation of genomic testing to diagnose rare diseases. Francisco has over 3 years of international experience in research, commercialisation and post-marketing surveillance of therapeutics and medical devices. He holds a Master's in Biotechnology Management, specialising in science commercialisation, and a Bsc. in Biotechnology Engineering with a research project in pharmacognosy.

These seminars are free. All are welcome to attend.
For more information contact the Centre for Health Policy CHP-enquiries@unimelb.edu.au / +61 3 8344 9111