3rd CIRCA Symposium 2020

PRE-educational EVENT

for junior doctors and trainees

Tuesday, 17 NOVEMBER 2020
17:00 – 19:30

17:00 INTRODUCTION
What is CIRCA (Clinical Immunogenomics Research Consortium Australasia)
Professor Stuart Tangye
Theme Leader, Immunity & Inflammation | Garvan Institute of Medical Research

17:10 Introduction to immunogenomics - Analysing and reporting genomes
Professor Leslie Burnett
Principal Medical Geneticist; Medical Director | Kinghorn Centre for Clinical Genomics

18:00 What doctors and patients need to know
Rebecca Macintosh
Genetic Counsellor, Centre for Clinical Genetics | Sydney Children's Hospital

18:30 -19:30 Overview of known genes and how to be involved in CIRCA
Professor Stuart Tangye
Theme Leader, Immunity & Inflammation | Garvan Institute of Medical Research

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Twitter: @CIRCA_genomics
Contact: Karen Enthoven k.enthoven@garvan.org.au

Background of speakers

Professor Stuart Tangye
Leader, Immunity & Inflammation Theme, Garvan; Head, Immunology & Immunodeficiency Lab, Garvan
Professor (Conjoint), St Vincent's Clinical School; Faculty of Medicine, UNSW Sydney Chair; International Union of Immunological Societies Expert Committee on Inborn Errors of Immunity.

I am an immunologist with expertise, and an established international reputation, in the fields of human immunology, immunodeficiency and inborn errors of immunity. I have worked at prestigious research institutes in the USA (DNAX Research Institute, Palo Alto, CA, USA; 1996-1999), and Australia (Centenary Institute, 2000-2006; Garvan Institute, 2006-present; Sydney, Aust.). I also undertook 6-month sabbatical study at the Rockefeller University (2015-2016; NY, USA). The overall goal of my research is to improve immunity in individuals with immune deficiencies, or during vaccination, or to attenuate immune responses in patients with autoimmune diseases. My research specifically aims to determine the requirements for the differentiation of B and T lymphocytes into effector cells that are necessary for the generation of protective immune responses following infection or vaccination. To achieve these goals, my lab studies cellular, biochemical and molecular biological processes in lymphocytes from healthy individuals as well as from patients with defined loss- or gain-of function mutations in specific genes that manifest as inborn errors of immunity resulting in primary immunodeficiencies (PIDs), autoimmunity, allergy and/or cancer. My studies have shed substantial light on mechanisms underlying compromised immunity in PIDs, thereby raising the prospect of improved therapies for individuals affected by such genetic mutations. As a result of these outputs, my lab is recognized as a leader in human immunology and immunodeficiency. In 2015, I established CIRCA, Australia's first integrated clinical program in immunogenomics, with clinical and research colleagues.

Rebecca Macintosh
Genetic Counsellor, Sydney Children's Hospital

Rebecca is currently employed in a full-time permanent position as a certified genetic counsellor at Sydney Children's Hospital, with a decade of experience. She works in the Clinical Genetics Department, seeing patients with a variety of conditions and liaising with multiple specialties across the hospital and with external providers.

Rebecca's particular interests are working with families with neurodevelopmental disorders and primary immunodeficiencies, and has had the opportunity to build and strengthen research relationships through her clinical work in both these areas which have benefited patient outcomes. Rebecca was granted a Sydney Children's Hospital Foundation starter grant in 2018 which assisted in the development of patient centred resources for parents of patients with severe genetic epilepsies.

Rebecca has been involved with Clinical Immunogenomics Research Consortium Australasia (CIRCA) since its inception. She initially providing genetic counselling to patients being investigated for an underlying cause for their primary immunodeficiency through the Sydney Children's Hospital immunogenomics clinic. Her involvement then expanded to additionally provide expertise on genomic consent, provision of results of exome/genome sequencing and as a member of the CIRCA Scientific Advisory Group, in additional to her clinical expertise.

Professor Leslie Burnett

Principal Medical Geneticist; Medical Director, Kinghorn Centre for Clinical Genomics
Conjoint Professor, St Vincent's Clinical School, UNSW;
Honorary Professor in Pathology and Genetic Medicine, Northern Clinical School, Faculty of Medicine and Health, The University of Sydney

Leslie is Principal Medical Geneticist and Medical Director within the Kinghorn Centre for Clinical Genomics at the Garvan Institute of Medical Research.
He is a Clinical Pathologist, with an unusually wide range of professional skills. He holds scope of practice in both Genetic Pathology and Chemical Pathology (Clinical Chemistry), and undertakes research in computational biology and bioinformatics.
He pioneered the development of a number of pathology and genetics initiatives, which are today mainstream clinical services. These include founding Australia's first Community Genetics program, being Medical Director of Australasia's first Whole Genome Sequencing laboratory, and developing the world's first pre- and post-analytical quality assurance programs in pathology. He has served as Ministerial appointee, Chairman, or President of a number of National and International bodies in pathology and genetics.
Leslie is Conjoint Professor at St Vincent's Clinical School, UNSW Medicine, Honorary Professor in Pathology and Genetic Medicine in the Northern Clinical School, Faculty of Medicine and Health, University of Sydney, and has been an Honorary Associate of the School of Information Technologies at the University of Sydney.
His current interests are in the areas of clinical genomics (especially in community and population screening), genetic pathology (especially in genomic pathology) and computational biology (especially in mechanisms of genomic regulation). He is a passionate teacher and communicator about the genetics and genomics revolution.