



# 08:30 Introduction to CIRCA and update on inborn errors of immunity

Prof Stuart Tangye

Theme Leader, Immunity & Inflammation, Garvan Institute of Medical Research

### 09:30 Dr Steven Holland

Director, Division of Intramural Research Chief, Immunopathogenesis Section National Institute of Allergy and Infectious Diseases, NIH, USA

# 10:30 How to find the needle... criteria for selection of monogenic immune diseases

Dr Paul Gray

Paediatric Immunologist, Sydney Children's Hospital Randwick (SCH) and Campbelltown Hospital Joint Head of Dept. of Immunology and Infectious Diseases SCH

## 11:30 Clinical immunogenomics and application to PID patients

Dr Melanie Wong

Paediatric Immunologist, Children's Hospital Westmead (CHW) Joint Head of Dept. of Immunology and Infectious Diseases CHW

## 12:00 The role of deep immunophenotyping in diagnosing PID

A/Prof Elissa Deenick

Laboratory Head | Lymphocyte Signalling and Activation Immunity & Inflammation Theme, Garvan Institute of Medical Research UNSW Scientia Fellow , Faculty of Medicine, UNSW Sydney

### A/Prof Cindy Ma

Laboratory Head | Immunodeficeincy & Immunology Immunity & Inflammation Theme, Garvan Institute of Medical Research

## 12:30 Mevalonate kinase deficiency and autoinflammation: insights

into disease mechanisms using new mouse avatars

Prof Mike Rogers

Head, Bone Therapeutics Laboratory, Healthy Aging, Theme, Garvan Institute of Medical Research

# 13:00 Immunity against SARS COV2 and COVID19 – lessons from inborn errors of immunity

Prof Stuart Tangye

Theme Leader, Immunity & Inflammation, Garvan Institute of Medical Research

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





#### 12:00 CVID-plus project and case presentation

Dr Ming Wei Lin

Adult Immunologist, Westmead Hospital,

Prof Tri Phan

St Vincent's Hospital and Garvan

#### 12:30 RNAseg and corporate partnerships - integrating genomics

and transcriptomics

A/Professor Sarah Kummerfeld

Head, Kinghorn Centre for Clinical Genomics (Scientific)

#### 13:00 Case Presentation

Dr Peter Hsu

Paediatric Immunologist, Children's Hospital Westmead

#### 13:30 Case presentation 4yo Caucasian girl hypogamma family

Dr Chaitanya Bodapati

Immunology and Allergy Fellow SCH

#### 14:30 Counselling challenges in the genomic age:

variable expressivity, polygenic vs monogenic, precision medicine, research findings vs clinically actionable findings

Prof Tri Phan (host) Clinician scientist, Garvan and St Vincent's Hospital Clinical Geneticists from RPAH:

Dr Lisa Worgan,

Dr Lisa Ewans

and Genetic Counsellor from SCH:

Rebecca Macintosh

#### Introduction to TNFAIP3 15:30

Prof Shane Grey Head, Transplant Immunology Lab, Garvan

#### 15:40 TNFAIP3 gene discussion

Prof Tri Phan (host) Clinician scientist, Garvan and St Vincent's Hospital Prof Shane Grey Head, Transplant Immunology Lab, Garvan

Nathan Zammit Postdoctoral Research Fellow at Garvan

Dr Paul Gray Paediatric Immunologist, Sydney Children's Hospital Randwick (SCH)

### Topics will include:

- diagnosing Polynesian and other non-Caucasian ethnic patients
- evolution and selective pressures on allelic frequencies
- animal models and disease mechanisms
- Four generation lupus family with interesting gene modifiers (eg TREX1)

#### 17:00 **Prof Isabelle Meyts**

Paediatric immunologist UZ Leuven Lead of Laboratory for Inborn Errors of Immunity, KU Leuven ESID President

#circasymposium2020 Twitter: @CIRCA\_genomics

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





08:50 Introduction

**Prof Stuart Tangye** 

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09:00 Overview of CTLA4 deficiency

Dr Gulbu Uzel

Laboratory of Clinical Immunology & Microbiology

National Institute of Allergy and Infectious Diseases, NIH, USA

10:00 The impact of genetic diagnosis on adult PID patients,

including therapeutics and transplantation

Dr Alisa Kane

Adult Immunologist, St Vincent's Hospital and Liverpool Hospital

Visiting Scientist at Garvan

10:30 Case Presentation: TB... or not PJP

A/Prof Allan Sturgess

Allergy Specialist & Immunologist, Pathologist (Immunologist), Rheumatologist

Head of Dept. of Rheumatology, St George Hospital

11:00 DOCK8 reversion

Dr Bethany Pillay

Research Officer, Immunodeficiency & Immunology Lab

Garvan Institute of Medical Research

11:30 CIRCA Collaborations and Projects and how to be involved

**Prof Stuart Tangye** 

-12:30 Theme Leader, Immunity & Inflammation,

Garvan Institute of Medical Research

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