

# Jeffrey Modell Diagnostic and Research Centers - Activity Report

Recommended "best practices" for Public Awareness, Physician Education and Research



## Center Information:

Date:	28 February 2019
Name of Center Director:	A/Prof. Menno C. van Zelm
Name of Institution:	JMF Centre Melbourne
City/Country:	Melbourne / Australia

## Progress and Updates:

### **Educational Activities:**

- PID Discussion group meetings
  - City-wide initiative of 3-4 evening sessions per year to discuss PID and difficult cases, aimed to educate clinical specialists, academics and registrars, exchange of expertise.
  - Three sessions held in 2018 with rotating hosts from JMF founding members (Royal Melbourne Hospital, Royal Children's Hospital, Alfred Health, Monash Health):  
19 April, July 26, October 11
  - Austin Health Haematology Dept joined the group in July 2018
- JMF Centre Melbourne Launch and Opening Symposium
  - 1-day event on 14 June 2018 with a scientific session, a clinical session, dedication ceremony and patient engagement session.
  - Attended by 80 delegates, including clinicians, scientists, patients and patient organisations from across Melbourne, Victoria and interstate (New South Wales and Australian Capital Territory)
- Sponsoring of PID breakfast session at annual meeting of the Australasian Society for Clinical Immunology and Allergy (ASCI) on 7 September 2018 (please see attached full programme, picture and news article on website <https://www.jmf-melbourne.org.au/news-events>)

### **Patient Support:**

- Interaction with Patient Organizations
  - Three patient organizations (IDFA, AusPIPS, Bloody Long Way) were invited and present at the opening symposium. IDFA presented and co-hosted the patient engagement session.
- Website
  - The Centre's website was launched in June 2018 and revamped in October 2018:  
<http://www.jmf-melbourne.org.au/>
  - Information on the Centre can be found here, and has been linked to a newly established Facebook page (<https://www.facebook.com/jmf.melbourne/>) to improve interactions with

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stakeholders (esp. JMF, and patient organisations). The website and Facebook page are aimed to:

- Promote activities
- Serve as portal to guide patients and professionals to the right resources and information

### Research:

#### ➤ Collaborative Studies

JMF Melbourne Exec member initiatives include local and Australia-wide genomic screening programs highlighted below.

#### Immunology Flagship Program of Melbourne Genomics Health Alliance

- JMF Exec members: Dr Vanessa Bryant, Dr Charlotte Slade, Dr Joanne Smart
- An Alliance of 10 leading healthcare and research organisations (including JMF Melbourne network Walter & Eliza Hall Institute, Royal Melbourne Hospital, Royal Children's Hospital, Monash Health).
- Dedicated to bringing global knowledge of genomics to benefit individual care of Victorians by demonstrating clinical utility and value of accredited clinical genomic sequencing for Victorians with suspected PID
- Completion of whole-exome sequencing, curation and reporting for 200 Victorians with suspected PID
- Preliminary findings include new diagnosis for 12% participants, changes to patient for 13% patients.
- Health economic analysis and QoL surveys now in progress.
- <https://www.melbournegenomics.org.au/news/providing-better-care-more-victorians-genomic-sequencing>

#### Genetic Immunology Flagship Program of Australian Genomic Health Alliance

- Flagship lead: Prof Matthew Cook, Centre for Personalised Immunology, ACT
- Members from JMF Melbourne exec committee: Dr Bosco, Dr, Bryant, Dr Slade, Dr Smart and A/Prof Van Zelm
- Aim to provide a national diagnostic and research network, a national approach to data federation and analysis, economic analysis and poly implications to the health system and build a genomic workforce, education and ethics.
- Recruitment of patients with suspected PID June -December 2019 for whole-genome sequencing.

#### Other Collaborative research studies between JMF Exec members:

- CD19 complex deficiencies: how do specific mutations lead to mild and severe clinical phenotypes?

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- Cohort analysis of adult patients with predominantly antibody deficiency in the State of Victoria (see publication below)
- Immunodeficiency and autoimmunity in patients with predominantly antibody deficiency
  
- Protocols Overview  
N/A
  
- Specific Aims
  - Providing a genetic diagnosis for patients with PID
  - Define the functional effects of pathogenic mutations
  - Understanding the contradictory presence of autoimmunity in patients with predominantly antibody deficiency
  
- Objectives Achieved
  - Multi-centre research database of adults with primary antibody deficiency
  - Retrospective analysis of diagnostic and clinical features of adult PAD patients
  
- Published Manuscripts
  - C.A. Slade, J.J. Bosco, T. Binh Giang, E. Kruse, R.G. Stirling, P.U. Cameron, F. Hore-Lacy, M.F. Sutherland, S.L. Barnes, S. Holdsworth, S. Ojaimi, G.A. Unglik, J. De Luca, M. Patel, K. Spriggs, Y. Tran, P. Auyeung, K. Nicholls, R.E. O'Hehir, P.D. Hodgkin, J.A. Douglass, V.L. Bryant, M.C. van Zelm (2018) Delayed diagnosis and complications of Predominantly Antibody Deficiencies in a cohort of Australian adults. *Frontiers Immunology*. 9(694):1-9.

### World Primary Immunodeficiency Week:

- Global Balloons & Bubbles Launch - Coordination with Local Patient Groups
- Media Outreach  
Herald Sun 14 June 2018: JMF Centre Melbourne launch
  
- Government Outreach
  - JMF Exec engagement with patient organisations (IDFA, AusPIPS) and Melbourne families affected by the absence of SCID newborn screening in Australia to discuss lobbying and discussions with State government Health Minister to implement SCID NBS in Victoria
  - Engage with patient organization, AusPIPS to establish working group for continued state-government funding and facilitation of SCIG access for PID patients in Australia.

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## Future Goals:

- Activities planned for next budget period

### Education / outreach

- Four PID Discussion group meetings in 2019: 21 March, 20 June, 12 September, 21 November
- Annual PID Update Day in April 2019 for Clinical specialists, registrars, laboratory scientists
- Continued engagement with patient organisations and families affected by the absence of SCID newborn screening in Australia to advocate for implementation of newborn screening for PIDs; for funding of SCIG in Victoria.
- JMF Melbourne is one of the stakeholders for a National Immunodeficiency Strategy for Australia. First meeting scheduled on March 8, 2019
- 2<sup>nd</sup> annual JMF Melbourne Immunodeficiency symposium to be held on Oct 11, 2019

### Diagnostics / research

- Pilot studies supplementing existing diagnostics, including:
  - Funding for genetic tests that are otherwise unavailable for 20 pediatric patients with suspected PID
  - Functional tests to supplement accredited tests (genetic diagnoses of uncertain significance) for diagnosis of PID.
  - Improve measurements for detection of impaired vaccination responses using neoantigens (e.g. Salmonella) or cell-based assays (collaboration with Prof. Sergio Rosenzweig, NIH, USA)- pilot
  - Extensive flow panels for to supplement diagnostics of selected cases

## Other:

### Attachments:

1. Programme ASCIA 2018 conference
2. Picture stage of PID breakfast session at ASCIA meeting showing JMF support
3. Programme National Immunodeficiency Strategy meeting, March 8, 2019