Primary Immunodeficiency (APID) Training Meeting 2019

PROGRAM

Dates: Sunday 26th May to Thursday 30th May 2019
Venue: Q Station, Manly NSW

International faculty: Dr Graham Davies, Dr Andrew Gennery, Dr Siobhan Burns
Local faculty: Dr Theresa Cole, Dr Paul Gray, A/Prof Katrina Randall, Prof Stuart Tangye, Dr Melanie Wong.

Delegates (24) listed in order of presentations in the program:

Dr Thérèse Boyle, Dr Josh Chatelier, Dr Carolyn Wijaya, Dr Matthew Krummenacher, Dr Lydia Chan, Dr Omega Leong, Dr Hannah Hu, Dr Yeri Ahn, Dr Katharine Foster, Dr Marsus Pumar, Dr Samantha Chan, Dr Michael Lane, Dr Helena Jang, Dr Wei-I Lee, Dr Ashisha Kallukaran, Dr Tim Brettig, Dr Timothy West, Dr Brittany Stevenson, Dr Sophie Willcocks, Dr Pei Dai, Dr Isabelle Bosi, Dr Jessica Lai, Dr Jacqueline Loprete, Dr Crystal Lam.

This program includes a case presentation by each delegate and each case has been allocated a total of 20 minutes, which comprises:

- 15 minutes for a slide presentation – whilst the number of slides isn’t limited, it is important that each presentation is completed within 15 minutes. The AV is optimized for PowerPoint slides in 16:9 ratio aspect.
- 5 minutes for questions and discussion.

This meeting is being held in conjunction with the ASCIA PID (APID) and TAPID Consultants meeting on 25-26th May 2019. There is no registration fee for these meetings due to the support received via an unrestricted educational grant from CSL Behring.

This is the 5th ASCIA PID Training meeting that has been held since 2007.

Participation in the ASCIA PID Advanced Training meetings is restricted to advanced trainees or recently qualified clinical immunology/allergy specialists, who have not previously attended these meetings. Delegate numbers are limited and preference is given to ASCIA members. It is our intention that ASCIA Associate (Trainee) members participate in at least one PID training meeting during advanced training, or soon after completion of advanced training. ASCIA Associate (Trainee) members are also encouraged to register and submit abstracts for ASCIA Annual Conferences, to further their knowledge in all areas of clinical immunology/allergy.

Sunday 26th May 2019 17:00 – 21:00 (A2 meeting room)

<table>
<thead>
<tr>
<th>Time</th>
<th>Activities</th>
<th>Speaker(s)</th>
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<tbody>
<tr>
<td>17:00 – 17:15</td>
<td>Registration</td>
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<tr>
<td>17:15 – 17:30</td>
<td>Welcome and introduction (15 min)</td>
<td>Dr Theresa Cole</td>
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<tr>
<td>17:30 – 18:30</td>
<td>Approach to the patient with recurrent infections</td>
<td>Dr Graham Davies</td>
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<td>Chair: Dr Theresa Cole</td>
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<tr>
<td>19:00 – 21:00</td>
<td>Dinner: Boilerhouse restaurant, Q Station</td>
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</table>
# Primary Immunodeficiency (APID) Training Meeting 2019

## PROGRAM

**Monday 27th May 2019 09:00 – 17:00 (A2 meeting room)**

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Chairs</th>
<th>Chair(s)</th>
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<tbody>
<tr>
<td>09:00 – 10:30</td>
<td>Session 1</td>
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<tr>
<td>09:00 – 09:30</td>
<td></td>
<td>Basic science of B cell disorders</td>
<td>Prof Stuart Tangye</td>
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<td>09:30 – 10:00</td>
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<td>THI / XLA</td>
<td>Dr Graham Davies</td>
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<tr>
<td>10:00 – 10:30</td>
<td></td>
<td>SCIG and IVIG</td>
<td>Dr Theresa Cole</td>
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<tr>
<td>10:30 – 11:00</td>
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<td>Morning Tea</td>
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<tr>
<td>11:00 – 12:30</td>
<td>Session 2</td>
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<tr>
<td>11:00 – 11:30</td>
<td></td>
<td>CVID</td>
<td>Dr Siobhan Burns</td>
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<tr>
<td>11:30 – 12:30</td>
<td>Case presentations:</td>
<td>CVID (common variable immune dysregulation)</td>
<td>Dr Thérèse Boyle</td>
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<tr>
<td>11:30 – 11:50</td>
<td></td>
<td>Complex patient with common variable immunodeficiency with multiple disease associated complications</td>
<td>Dr Josh Chatelier</td>
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<td>11:50 – 12:10</td>
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<td>A young woman with enlarged lymph nodes</td>
<td>Dr Carolyn Wijaya</td>
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<tr>
<td>12:10 – 12:30</td>
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<td>Lunch: Views restaurant</td>
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<tr>
<td>13:30 – 15:00</td>
<td>Session 3</td>
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<tr>
<td>13:30 – 14:00</td>
<td></td>
<td>Hyper-IgM syndromes (X-linked, AR, NEMO)</td>
<td>Dr Andrew Gennery</td>
</tr>
<tr>
<td>14:00 – 15:00</td>
<td>Case presentations:</td>
<td>Clinical spectrum of activated PI3 kinase delta syndrome (APDS)</td>
<td>Dr Matthew Krummenacher</td>
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<tr>
<td>14:00 – 14:20</td>
<td></td>
<td>Activated PI3K delta syndrome type</td>
<td>Dr Lydia Chan</td>
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<tr>
<td>14:20 – 15:00</td>
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<td>Discussion</td>
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<td>15:00 – 15:30</td>
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<td>Afternoon Tea</td>
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<tr>
<td>15:30 – 17:00</td>
<td>Session 4</td>
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<tr>
<td>15:30 – 16:00</td>
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<td>Thymic disorders and DiGeorge syndrome</td>
<td>Dr Graham Davies</td>
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<td>16:00 – 16:30</td>
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<td>DNA repair defects and ataxia telangiectasia</td>
<td>Dr Andrew Gennery</td>
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<tr>
<td>16:30 – 16:50</td>
<td>Case presentation:</td>
<td>Refractory cutaneous and visceral granulomatous disease in ataxia telangiectasia</td>
<td>Dr Omega Leong</td>
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<tr>
<td>16:50 – 17:00</td>
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<td>Discussion</td>
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<tr>
<td>19:00 – 21:00</td>
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<td>Dinner: Hugo’s restaurant, Manly Wharf (minibus transfers provided)</td>
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# Primary Immunodeficiency (APID) Training Meeting 2019

**PROGRAM**

**Tuesday 28th May 2019 09:00 – 17:00 (A2 meeting room)**

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<thead>
<tr>
<th>Time</th>
<th>Session 1</th>
<th>Chairs:</th>
<th>Speakers</th>
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</thead>
<tbody>
<tr>
<td>09:00 – 10:30</td>
<td>Basic science of combined immunodeficiencies</td>
<td>Dr Paul Gray, Dr Melanie Wong</td>
<td>Prof Stuart Tangye</td>
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<tr>
<td>09:00 – 09:30</td>
<td>SCID cases, pre HSCT management and transplant</td>
<td>Dr Theresa Cole</td>
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<tr>
<td>09:30 – 10:00</td>
<td>Non-SCID HSCT</td>
<td>Dr Andrew Gennery</td>
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<tr>
<td>10:00 – 10:30</td>
<td>Morning Tea</td>
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<tr>
<th>Time</th>
<th>Session 2</th>
<th>Chairs:</th>
<th>Speakers</th>
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</thead>
<tbody>
<tr>
<td>11:00 – 12:30</td>
<td>Cellular and gene therapy</td>
<td>Dr Graham Davies</td>
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<tr>
<td>11:00 – 11:30</td>
<td>Wiskott- Aldrich syndrome and related disorders</td>
<td>Dr Siobhan Burns</td>
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<tr>
<td>11:30 – 12:00</td>
<td>Case presentation: ADA2 deficiency - monogenic polyarteritis with hypogammaglobulinaemia</td>
<td>Dr Hannah Hu</td>
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<tr>
<td>12:00 – 12:30</td>
<td>Discussion</td>
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<tr>
<td>12:30 – 13:30</td>
<td>Lunch: Views restaurant</td>
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<thead>
<tr>
<th>Time</th>
<th>Session 3</th>
<th>Chairs:</th>
<th>Speakers</th>
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</thead>
<tbody>
<tr>
<td>13:30 – 15:00</td>
<td>Neutrophil disorders and chronic granulomatous disease (CGD): presentation, diagnosis, management and complications</td>
<td>Dr Siobhan Burns</td>
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<tr>
<td>13:30 – 14:00</td>
<td>Discerning variants</td>
<td></td>
<td>Dr Yeri Ahn</td>
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<tr>
<td>14:00 – 14:20</td>
<td>Chronic granulomatous disease (CGD) presenting in adulthood</td>
<td>Dr Katharine Foster</td>
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<tr>
<td>14:20 – 14:40</td>
<td>A case of invasive aspergillus felis infection with thyroid, liver and splenic aspergillus abscesses in a patient with chronic granulomatous disease (CGD)</td>
<td>Dr Marsus Pumar</td>
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<td>14:40 – 15:00</td>
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<tr>
<td>15:00 – 15:30</td>
<td>Afternoon Tea</td>
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<tr>
<th>Time</th>
<th>Session 4</th>
<th>Chairs:</th>
<th>Speakers</th>
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</thead>
<tbody>
<tr>
<td>15:30 – 17:00</td>
<td>PIDs associated with hyper IgE (including STAT3, DOCK8, ZNF341)</td>
<td>Dr Paul Gray, Dr Graham Davies</td>
<td>Prof Stuart Tangye</td>
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<tr>
<td>15:30 – 16:00</td>
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<tr>
<td>16:00 – 16:20</td>
<td>A case of non-eosinophilic myocarditis in AD-HIES</td>
<td>Dr Samantha Chan</td>
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<tr>
<td>16:20 – 16:40</td>
<td>Novel STAT3 mutation and hyper IgE syndrome</td>
<td>Dr Michael Lane</td>
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<tr>
<td>16:40 – 17:00</td>
<td>A job worth the effort</td>
<td>Dr Helena Jang</td>
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<td>16:00 – 17:00</td>
<td>Case presentations:</td>
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<tr>
<td>19:00 – 21:00</td>
<td>Dinner: Garfish restaurant, Manly (minibus transfers provided)</td>
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**Primary Immunodeficiency (APID) Training Meeting 2019**

**PROGRAM**

**Wednesday 29th May 2019 09:00 – 17:00 (A2 meeting room)**

<table>
<thead>
<tr>
<th>Time</th>
<th>Session</th>
<th>Chairs</th>
<th>Presenters</th>
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</thead>
<tbody>
<tr>
<td>09:00 – 10:30</td>
<td>Session 1</td>
<td>Dr Andrew Gennery, Dr Melanie Wong</td>
<td>Prof Stuart Tangye</td>
</tr>
<tr>
<td>09:00 – 09:30</td>
<td>Basic science of immune dysregulation</td>
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<tr>
<td>09:30 – 10:00</td>
<td>ALPS and other PID associated cytopenias</td>
<td>A/Prof Katrina Randall</td>
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<tr>
<td>10:00 – 10:20</td>
<td>Case presentation</td>
<td>PID presented initially with pancytopenia and micro-nutrient deficiency</td>
<td>Dr Wei-I Lee</td>
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<tr>
<td>10:20 – 10:30</td>
<td>Discussion</td>
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<tr>
<td>10:30 – 11:00</td>
<td>Morning Tea</td>
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<tr>
<td>11:00 – 12:30</td>
<td>Session 2</td>
<td>Dr Theresa Cole, A/Prof Katrina Randall</td>
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<tr>
<td>11:00 – 11:30</td>
<td>Early onset IBD</td>
<td>Dr Melanie Wong</td>
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<tr>
<td>11:30 – 12:00</td>
<td>CTLA/LRB</td>
<td>Dr Paul Gray</td>
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<tr>
<td>12:00 – 12:20</td>
<td>Case presentation</td>
<td>Could this be evolving CGD?</td>
<td>Dr Ashisha Kallukaran</td>
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<td>12:20 – 12:30</td>
<td>Discussion</td>
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<tr>
<td>12:30 – 13:30</td>
<td>Lunch: Views restaurant</td>
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<tr>
<td>13:30 – 15:00</td>
<td>Session 3</td>
<td>Dr Siobhan Burns, Dr Paul Gray</td>
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<tr>
<td>13:30 – 14:00</td>
<td>Familial HLH including XLP</td>
<td>Dr Melanie Wong</td>
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<tr>
<td>14:00 – 14:00</td>
<td>Case presentations:</td>
<td>Dr Tim Brettig</td>
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<tr>
<td>14:00 – 14:20</td>
<td>Recurrent fevers</td>
<td>Dr Timothy West</td>
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<tr>
<td>14:20 – 14:40</td>
<td>HLH in an adult male with RAS-associated autoimmune lymphoproliferative syndrome (RALD)</td>
<td>Dr Theresa Cole</td>
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<tr>
<td>14:40 – 15:00</td>
<td>HSCT for HLH</td>
<td>Dr Theresa Cole</td>
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<tr>
<td>15:00 – 15:30</td>
<td>Afternoon Tea</td>
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<tr>
<td>15:30 – 17:00</td>
<td>Session 4</td>
<td>Prof Stuart Tangye, Dr Melanie Wong</td>
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<tr>
<td>15:30 – 16:00</td>
<td>Autoinflammatory conditions</td>
<td>Dr Paul Gray</td>
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<td>16:00 – 17:00</td>
<td>Case presentations:</td>
<td>Dr Brittany Stevenson</td>
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<td>16:00 – 16:20</td>
<td>Ruxolitinib in STING associated vasculopathy with onset in infancy (SAVI)</td>
<td>Dr Sophie Willcocks</td>
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<td>16:20 – 16:40</td>
<td>Well PLAID</td>
<td>Dr Pei Dai</td>
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<td>16:40 – 17:00</td>
<td>Getting High on Zinc</td>
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<tr>
<td>19:00 – 21:00</td>
<td>Dinner: Boilerhouse restaurant, Q Station</td>
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# Primary Immunodeficiency (APID) Training Meeting 2019

## PROGRAM

**Thursday 30th May 2019 08:30 – 16:30 (P16 meeting room)**

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<th>Session 1</th>
<th>Chairs: Dr Theresa Cole, Dr Andrew Gennery</th>
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<tbody>
<tr>
<td>08:30 – 10:00</td>
<td>Genomic diagnosis in PID</td>
<td>Dr Paul Gray</td>
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<tr>
<td>08:30 – 09:00</td>
<td>Mucocutaneous candidiasis</td>
<td>A/Prof Katrina Randall</td>
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<tr>
<td>09:00 – 09:30</td>
<td>TLR pathway defects (including TLR3, IRAK4, MyD88)</td>
<td>A/Prof Katrina Randall</td>
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<td>09:30 – 10:00</td>
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<tr>
<th>Time</th>
<th>Session 2</th>
<th>Chairs: A/Prof Katrina Randall, Dr Melanie Wong</th>
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<tbody>
<tr>
<td>10:30 – 12:00</td>
<td>MSMD</td>
<td>Dr Siobhan Burns</td>
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<tr>
<td>11:00 – 12:00</td>
<td><strong>Case presentations:</strong></td>
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<tr>
<td>11:00 – 11:20</td>
<td>Disseminated non-tuberculous mycobacteria and the evaluation of mendelian susceptibility to mycobacterial diseases (MSMD)</td>
<td>Dr Isabelle Bosi</td>
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<tr>
<td>11:20 – 11:40</td>
<td>A case of MSMD</td>
<td>Dr Jessica Lai</td>
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<tr>
<td>11:40 – 12:00</td>
<td>Diagnosis of stat1 gain of function disorder in an adult</td>
<td>Dr Jacqueline Loprete</td>
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<tr>
<td>12:00 – 13:00</td>
<td>Lunch: P16 Balcony</td>
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<thead>
<tr>
<th>Time</th>
<th>Session 3</th>
<th>Chairs: Dr Theresa Cole, Prof Stuart Tangye</th>
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<tbody>
<tr>
<td>13:00 – 13:40</td>
<td>Complement deficiencies, including hereditary angioedema (HAE)</td>
<td>Dr Melanie Wong</td>
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<tr>
<td>13:40 – 14:00</td>
<td><strong>Case presentation:</strong></td>
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<tr>
<td>13:40 – 14:00</td>
<td>The curious diagnosis of hereditary angioedema (HAE) in systemic lupus erythematosus (SLE) with congenital C4 deficiency</td>
<td>Dr Crystal Lam</td>
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<tr>
<td>14:00 – 14:30</td>
<td>Advances in PID diagnosis and treatment</td>
<td>Dr Andrew Gennery</td>
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<tr>
<td>14:30 – 15:00</td>
<td>Afternoon Tea</td>
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<thead>
<tr>
<th>Time</th>
<th>Session 4</th>
<th>Chairs: Dr Theresa Cole, Dr Melanie Wong</th>
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<tbody>
<tr>
<td>15:00 – 16:00</td>
<td>Panel discussion / Q&amp;A</td>
<td>All Faculty</td>
</tr>
<tr>
<td>16:00 – 16:30</td>
<td>Thank you and closing</td>
<td>Dr Theresa Cole, Dr Melanie Wong</td>
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