## Thursday 30 August

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<thead>
<tr>
<th>Time</th>
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| 8.30am - 8.45am | **Overview and the Rare Brain Disease Research Community**  
Chaired by Emma Scotter (University of Auckland)  
Welcome and introduction: Emma Scotter sponsored by Thermo Fisher Scientific  
University of Auckland | Dart Room, QT Building                                                             |
| 8.45am – 9.00am | Cathy Stinear  
Neurological Foundation Scientific Advisory Committee Chair  
*Bird’s eye view of rare brain disease research in NZ* | Dart Room, QT Building                                                             |
| 9.00am - 9.15am | Cliff Abraham sponsored by Bio-Strategy  
Brain Research New Zealand  
BRNZ and rare brain diseases | Dart Room, QT Building                                                             |
| 9.15am – 9.30am | Beth Watson (R1)  
MND New Zealand  
*From support to research – or are they intertwined?* | Dart Room, QT Building                                                             |
| 9.30am – 9.50am | Buzz Burrell (R2)  
University of Otago  
*Kearns Sayre syndrome – a Dad’s Perspective* | Dart Room, QT Building                                                             |
| 9.50am-10.30am | Morning Tea | Trades Area                                                                      |
| 10.30am-10.50am | Nicola Swain (R3)  
Southern Health and Disability Ethics Committee  
The Insider’s guide to NZ Ethics approval - Pitfalls and tips. | Dart Room, QT Building                                                             |
| 10.50am –11.10am | Collette Bromhead (R4)  
NZORD  
*Rare brain diseases policy and treatment funding* | Dart Room, QT Building                                                             |
| 11.10am-11.40am | Caroline De Luca & Rebecca Elliott (R5)  
PHARMAC  
*Funding medicine for rare disorders: PHARMAC’s revised policy settings and processes* | Dart Room, QT Building                                                             |
| 11.40-12.00pm  | Ronald Melki (R6)  
Paris-Saclay Institute of Neuroscience  
Protein Aggregation and templating | Dart Room, QT Building                                                             |
| 12:00-12:20pm  | Pritika Narayan (R7)  
University of Auckland  
*DNA and histone methylation changes in Huntington’s disease* | Dart Room, QT Building                                                             |
| 12.20pm- 12.30pm | All session speakers  
*Question Panel* | Dart Room, QT Building                                                             |
<p>| 12.30pm-1.30pm | Lunch | Trades Area                                                                      |</p>
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| 1.30pm – 1.50pm   | Bradley Turner (R8)           | *sponsored by Thermo Fisher Scientific*  
Florey Institute  
*Molecular mechanisms of MND/SMA* | Dart Room, QT Building               |
| 1.50pm-2.10pm     | Mirella Dottori (R9)          | University of Melbourne  
*Developing Regenerative Therapies for Friedreich’s Ataxia using Sensory Neurons derived from induced Pluripotent Stem Cells* | Dart Room, QT Building |
| 2.10pm-2.30pm     | Andrew Munkacsi (R10)         | Victoria University  
*The power of the yeast model of Niemann-Pick Type C disease from fundamental sterol transport to rare disease clinical trials* | Dart Room, QT Building |
| 2.30pm-2.45pm     | Grace Chen (R11)              | Massey University  
*Occupation and Motor Neurone Disease: A New Zealand Case-Control Study* | Dart Room, QT Building |
| 2.45pm-3:00pm     | Samantha Murray (R12)         | University of Otago  
*Ocular gene therapy in ovine models of CLN5 and CLN6 Batten disease* | Dart Room, QT Building |
| 3.00-3:30pm       | All session speakers          | Question Panel                                                                | Dart Room, QT Building |
| 3.30pm-4.00pm     |                                | Afternoon Tea                                                                  | Trades Area            |
| 4.00pm-4.20pm     | Louise Bicknell (R13)         | University of Otago  
*Gene hunting in Meier-Gorlin Syndrome* | Dart Room, QT Building               |
| 4.20pm-4.40pm     | Richard Roxburgh (R14)        | Auckland District Health Board  
*Gene hunting in CANVAS Syndrome* | Dart Room, QT Building               |
| 4.40pm-5.00pm     | Courtney Forgeng (R15)        | Genetic Health Service NZ  
*Genetic counseling in the genomic era* | Dart Room, QT Building               |
| 5.00pm-5.20pm     | Anjali Henders (R16)          | *sponsored by Thermo Fisher Scientific*  
University of Queensland  
*The Ice Bucket Challenge Sporadic ALS Australia Systems Genomics Consortium: SALSA-SGC* | Dart Room, QT Building               |
| 5.20pm-5.40pm     | All session speakers          | Question Panel                                                                | Dart Room, QT Building |
| 5.40pm-6.40pm     |                                | *Poster session*  
Sponsored by NZORD  
Judges: Collette Bromhead and Stephanie Hughes  
*Social Mixer with drinks and nibbles*  
*sponsored by Merck & pHScientific* | Marquee  
Trades Area |

**Molecular Mechanisms cont.**  
*Chaired by Stephanie Hughes (University of Otago)*

**Gene Hunting**  
*Chaired by Chris Shaw (King’s College London)*

**Rare Genetic Diseases of the Brain Dinner – Blue Kanu 7pm**
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<tr>
<td>8.30am - 8.40am</td>
<td><strong>Day 2 Welcome and overview</strong> Jayne McLean</td>
<td>Dart Room, QT Building</td>
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| 8.40am – 9.00am | Maurice Curtis (R17)  
University of Auckland  
*Neurological Foundation Douglas Human Brain Bank* | Dart Room, QT Building            |
| 9.00am – 9.20am | Mike Dragunow (R18) *sponsored by Bio-Strategy*  
University of Auckland  
Hugh Green BioBank | Dart Room, QT Building            |
| 9.20am – 9.30am | Kerry Walker (R19)  
Auckland District Health Board  
*The New Zealand Motor Neurone Disease Registry; One Year On* | Dart Room, QT Building            |
| 9.30am – 9.40am | Jayne McLean (R20)  
University of Auckland  
*New Zealand Motor Neuron Disease Research Network* | Dart Room, QT Building            |
| 9.40-10.00am   | **All session speakers**  
*Question Panel* | Dart Room, QT Building            |
| 10.00am-10.30am | **Morning Tea** | Trades Area                      |

**Genetic Disease Models and Genetics Oversight NZ (joint with Applied Genetic Technologies)**  
Chair Francis Hunter, University of Auckland

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| 10.30am-10.50am | Tracey Dickson (R21)  
University of Tasmania  
*Getting the Balance Right: Targeting Excitatory Dysfunction in the ALS Cortex* | Clancy’s Room           |
| 10.50am –11.10am | Imke Tammen (R22)  
University of Sydney  
*Generation of a sheep model of CLN7 Batten disease using the CRISPR/Cas9 genome editing system – preliminary results and ethical reflections* | Clancy’s Room           |
| 11.10am-11.30am | Renee Handley (R23)  
University of Auckland  
*Brain urea increase is an early pathological event in Huntington’s disease* | Clancy’s Room           |
| 11.30am-11.50pm | Peter Dearden (R24)  
Genomics Aotearoa, University of Otago  
*Genomics Aotearoa: Aiming to improve the use of genomics in New Zealand* | Clancy’s Room           |
| 11.50am- 12.10pm | Kate Neas (R25)  
Genetic Health Service New Zealand  
*A step towards unifying health genomic activity in NZ  
Genomic Health Alliance New Zealand (GHANZ)* | Clancy’s Room           |
| 12.10pm-12.30pm | **Discussion session**  
*Sharing/improving/linking research resources* | Clancy’s Room           |
<p>| 12.30pm-1.30pm | <strong>Lunch</strong> | Trades Area                      |</p>
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<td>1.30pm – 1.50pm</td>
<td>Nadia Mitchell (R26)</td>
<td>Efficacy of gene therapy in a sheep model of CLN5 Batten disease</td>
<td>Dart Room, QT Building</td>
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<tr>
<td>1.50-2.10pm</td>
<td>Miriam Rodrigues (R27)</td>
<td>Spinal Muscular Atrophy:Treating the Cause</td>
<td>Dart Room, QT Building</td>
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<tr>
<td>2.10-3.00pm</td>
<td>Prof. Chris Shaw (R28)</td>
<td>Moving from Gene Discovery to Gene Therapy for Motor Neuron Disease and Frontotemporal Dementia</td>
<td>Dart Room, QT Building</td>
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<tr>
<td>3.00-3.15pm</td>
<td>All session speakers</td>
<td>Question Panel</td>
<td>Dart Room, QT Building</td>
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<td>3.15pm-3.30pm</td>
<td>Meeting close</td>
<td>Emma Scotter</td>
<td>Dart Room, QT Building</td>
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<td>3.30pm-4.30pm</td>
<td>MND ROUND TABLE (all stakeholders)</td>
<td>Research directions</td>
<td>Dart Room, QT Building</td>
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We gratefully acknowledge the support of our meeting sponsors: