

## QRW Programme

**Rare Genetic Diseases of the Brain**  
Thursday 30 August – Friday 31 August, 2018  
Rydges Hotel, Queenstown, New Zealand

| <b>Thursday 30 August</b>  |  |                        |
|--|--|------------------------|
| Time   | Details  | Location               |
| <b>Overview and the Rare Brain Disease Research Community</b><br><b>Chaired by Emma Scotter (University of Auckland)</b> |  |                        |
| 8.30am - 8.45am<br>15 mins   | <b>Welcome and introduction: Emma Scotter</b> <i>sponsored by Thermo Fisher Scientific</i><br>University of Auckland   | Dart Room, QT Building |
| 8.45am – 9.00am<br>15 mins   | <b>Cathy Stinear</b><br>Neurological Foundation Scientific Advisory Committee Chair<br><i>Bird's eye view of rare brain disease research in NZ</i>           | Dart Room, QT Building |
| 9.00am - 9.15am<br>15 mins   | <b>Cliff Abraham</b> <i>sponsored by Bio-Strategy</i><br>Brain Research New Zealand<br><i>BRNZ and rare brain diseases</i>                                   | Dart Room, QT Building |
| 9.15am – 9.30am<br>15 mins   | <b>Beth Watson (R1)</b><br>MND New Zealand<br><i>From support to research – or are they intertwined?</i>   | Dart Room, QT Building |
| 9.30am – 9.50am<br>20 mins   | <b>Buzz Burrell (R2)</b><br>University of Otago<br><i>Kearns Sayre syndrome – a Dad's Perspective</i>  | Dart Room, QT Building |
| 9.50am-10.30am<br>40 mins  | <b>Morning Tea</b>   | Trades Area            |
| <b>Policy, Ethics and Funding</b><br><b>Chaired by Miriam Rodrigues (Auckland DHB)</b>                                   |  |                        |
| 10.30am-10.50am<br>20 mins   | <b>Nicola Swain (R3)</b><br>Southern Health and Disability Ethics Committee<br><i>The Insider's guide to NZ Ethics approval - Pitfalls and tips.</i>         | Dart Room, QT Building |
| 10.50am –11.10am<br>20 mins  | <b>Collette Bromhead (R4)</b><br>NZORD<br><i>Rare brain diseases policy and treatment funding</i>  | Dart Room, QT Building |
| 11.10am-11.40am<br>30 mins   | <b>Caroline De Luca &amp; Rebecca Elliott (R5)</b><br>PHARMAC<br><i>Funding medicine for rare disorders: PHARMAC's revised policy settings and processes</i> | Dart Room, QT Building |
| <b>Molecular Mechanisms</b><br><b>Chaired by Stephanie Hughes (University of Otago)</b>                                  |  |                        |
| 11.40-12.00pm<br>20 mins   | <b>Ronald Melki (R6)</b><br>Paris-Saclay Institute of Neuroscience<br><i>Protein Aggregation and templating</i>  | Dart Room, QT Building |
| 12:00-12:20pm<br>20 mins   | <b>Pritika Narayan (R7)</b><br>University of Auckland<br><i>DNA and histone methylation changes in Huntington's disease</i>                                  | Dart Room, QT Building |
| 12.20pm- 12.30pm<br>30 mins  | <b>All session speakers</b><br><i>Question Panel</i>   | Dart Room, QT Building |
| 12.30pm-1.30pm   | <b>Lunch</b>   | Trades Area            |

| <b>Molecular Mechanisms cont.</b><br><b>Chaired by Stephanie Hughes (University of Otago)</b> |   |  |
|---|---|--|
| 1.30pm – 1.50pm<br>20 mins  | <b>Bradley Turner (R8)</b> <i>sponsored by Thermo Fisher Scientific</i><br>Florey Institute<br><i>Molecular mechanisms of MND/SMA</i>   | <b>Dart Room, QT Building</b>            |
| 1.50pm-2.10pm<br>20 mins  | <b>Mirella Dottori (R9)</b><br>University of Melbourne<br><i>Developing Regenerative Therapies for Friedreich's Ataxia using Sensory Neurons derived from induced Pluripotent Stem Cells</i>            | <b>Dart Room, QT Building</b>            |
| 2.10pm-2.30pm<br>20 mins  | <b>Andrew Munkacsi (R10)</b><br>Victoria University<br><i>The power of the yeast model of Niemann-Pick Type C disease from fundamental sterol transport to rare disease clinical trials</i>             | <b>Dart Room, QT Building</b>            |
| <b>Short talks</b>  |   |  |
| 2.30pm-2.45pm<br>15 mins  | <b>Grace Chen (R11)</b><br>Massey University<br><i>Occupation and Motor Neurone Disease: A New Zealand Case-Control Study</i>   | <b>Dart Room, QT Building</b>            |
| 2.45pm-3:00pm<br>15 mins  | <b>Samantha Murray (R12)</b><br>University of Otago<br><i>Ocular gene therapy in ovine models of CLN5 and CLN6 Batten disease</i>   | <b>Dart Room, QT Building</b>            |
| 3.00-3:30pm   | <b>All session speakers</b><br><i>Question Panel</i>  | <b>Dart Room, QT Building</b>            |
| 3.30pm-4.00pm   | <b>Afternoon Tea</b>  | <b>Trades Area</b>                       |
| <b>Gene Hunting</b><br><b>Chaired by Chris Shaw (King's College London)</b>                   |   |  |
| 4.00pm-4.20pm<br>20 mins  | <b>Louise Bicknell (R13)</b><br>University of Otago<br><i>Gene hunting in Meier-Gorlin Syndrome</i>   | <b>Dart Room, QT Building</b>            |
| 4.20pm-4.40pm<br>20 mins  | <b>Richard Roxburgh (R14)</b><br>Auckland District Health Board<br><i>Gene hunting in CANVAS Syndrome</i>   | <b>Dart Room, QT Building</b>            |
| 4.40pm-5.00pm<br>20 mins  | <b>Courtney Forgeng (R15)</b><br>Genetic Health Service NZ<br><i>Genetic counseling in the genomic era</i>  | <b>Dart Room, QT Building</b>            |
| 5.00pm-5.20pm<br>20 mins  | <b>Anjali Henders (R16)</b> <i>sponsored by Thermo Fisher Scientific</i><br>University of Queensland<br><i>The Ice Bucket Challenge Sporadic ALS Australia Systems Genomics Consortium: SALSA -SGC</i>  | <b>Dart Room, QT Building</b>            |
| 5.20pm-5.40pm<br>20 mins  | <b>All session speakers</b><br><i>Question Panel</i>  | <b>Dart Room, QT Building</b>            |
| 5.40pm-6.40pm<br>60 mins  | <b>Poster session</b><br>Sponsored by NZORD<br>Judges: Collette Bromhead and Stephanie Hughes<br><b>Social Mixer with drinks and nibbles</b> <i>sponsored by sponsored by Merck &amp; pH Scientific</i> | <b>Marquee</b><br><br><b>Trades Area</b> |
| <b>Rare Genetic Diseases of the Brain Dinner – Blue Kanu 7pm</b>                              |   |  |

## Friday 31 August

| Time   | Details   | Location               |
|--|---|------------------------|
| <b>Research Resources</b><br><b>Chaired by Ruth Empson, University of Otago</b>  |   |                        |
| 8.30am - 8.40am<br>10 mins   | <b>Day 2 Welcome and overview: Jayne McLean</b><br>University of Auckland   | Dart Room, QT Building |
| 8.40am – 9.00am<br>20 mins   | <b>Maurice Curtis (R17)</b><br>University of Auckland<br><i>Neurological Foundation Douglas Human Brain Bank</i>  | Dart Room, QT Building |
| 9.00am – 9.20am<br>20 mins   | <b>Mike Dragunow (R18)</b> <i>sponsored by Bio-Strategy</i><br>University of Auckland<br><i>Hugh Green BioBank</i>  | Dart Room, QT Building |
| 9.20am – 9.30am<br>10 mins   | <b>Kerry Walker (R19)</b><br>Auckland District Health Board<br><i>The New Zealand Motor Neurone Disease Registry; One Year On</i>   | Dart Room, QT Building |
| 9.30am – 9.40am<br>10 mins   | <b>Jayne McLean (R20)</b><br>University of Auckland<br><i>New Zealand Motor Neuron Disease Research Network</i>   | Dart Room, QT Building |
| 9.40-10.00am<br>20 mins  | <b>All session speakers</b><br><i>Question Panel</i>  | Dart Room, QT Building |
| 10.00am-10.30am  | <b>Morning Tea</b>  | Trades Area            |
| <b>Genetic Disease Models and Genetics Oversight NZ (joint with Applied Genetic Technologies)</b><br><b>Chair Francis Hunter, University of Auckland</b> |   |                        |
| 10.30am-10.50am<br>20 mins   | <b>Tracey Dickson (R21)</b><br>University of Tasmania<br><i>Getting the Balance Right: Targeting Excitatory Dysfunction in the ALS Cortex</i>   | Clancy's Room          |
| 10.50am –11.10am<br>20 mins  | <b>Imke Tammen (R22)</b><br>University of Sydney<br><i>Generation of a sheep model of CLN7 Batten disease using the CRISPR/Cas9 genome editing system – preliminary results and ethical reflections</i> | Clancy's Room          |
| 11.10am-11.30am<br>20 mins   | <b>Renee Handley (R23)</b><br>University of Auckland<br><i>Brain urea increase is an early pathological event in Huntington's disease</i>   | Clancy's Room          |
| 11.30am-11.50pm<br>20 mins   | <b>Peter Dearden (R24)</b><br>Genomics Aotearoa, University of Otago<br><i>Genomics Aotearoa: Aiming to improve the use of genomics in New Zealand</i>  | Clancy's Room          |
| 11.50am- 12.10pm<br>20 mins  | <b>Kate Neas (R25)</b><br>Genetic Health Service New Zealand<br><i>A step towards unifying health genomic activity in NZ Genomic Health Alliance New Zealand (GHANZ)</i>                                | Clancy's Room          |
| 12.10pm-12.30pm<br>20 mins   | <b>Discussion session</b><br><i>Sharing/improving/linking research resources</i>  | Clancy's Room          |
| 12.30pm-1.30pm   | <b>Lunch</b>  | Trades Area            |

| <b>Emerging Therapies and keynote</b><br><b>Chaired by Prof. Anthony Hannan, University of Melbourne</b> |  |                               |
|--|--|-------------------------------|
| 1.30pm – 1.50pm<br>20 mins   | <b>Nadia Mitchell (R26)</b><br>University of Otago<br><i>Efficacy of gene therapy in a sheep model of CLN5 Batten disease</i>  | <b>Dart Room, QT Building</b> |
| 1.50-2.10pm<br>20 mins   | <b>Miriam Rodrigues (R27)</b><br>Auckland District Health Board<br><i>Spinal Muscular Atrophy: Treating the Cause</i>  | <b>Dart Room, QT Building</b> |
| 2.10-3.00pm<br>50 mins   | <b>Prof. Chris Shaw (R28)</b> <i>sponsored by In Vitro Technologies</i><br>King's College London<br><i>Moving from Gene Discovery to Gene Therapy for Motor Neuron Disease and Frontotemporal Dementia</i> | <b>Dart Room, QT Building</b> |
| 3.00-3.15pm<br>15 mins   | <b>All session speakers</b><br><i>Question Panel</i>   | <b>Dart Room, QT Building</b> |
| 3.15pm-3.30pm<br>15 mins   | <b>Meeting close</b><br>Emma Scotter<br>University of Auckland   | <b>Dart Room, QT Building</b> |
| 3.30pm-4.30pm<br>60 mins   | <b>MND ROUND TABLE (all stakeholders)</b><br><i>Research directions</i>  | <b>Dart Room, QT Building</b> |

**We gratefully acknowledge the support of our meeting sponsors:**

