

## QRW Programme

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

### Thursday 30 August

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

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

## Friday 31 August

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

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