

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

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

Friday 31 August		
Time	Details	Location
Research Resources Chaired by Ruth Empson, University of Otago		
8.30am - 8.40am 10 mins	Day 2 Welcome and overview : Jayne McLean University of Auckland	
8.40am – 9.00am 20 mins	Maurice Curtis (R17) University of Auckland <i>Neurological Foundation Douglas Human Brain Bank</i>	
9.00am – 9.20am 20 mins	Mike Dragunow (R18) University of Auckland <i>Hugh Green BioBank</i>	
9.20am – 9.30am 10 mins	Kerry Walker (R19) Auckland District Health Board <i>The New Zealand Motor Neurone Disease Registry; One Year On</i>	
9.30am – 9.40am 10 mins	Jayne McLean (R20) University of Auckland <i>New Zealand Motor Neuron Disease Research Network</i>	
9.40-10 20 mins	All session speakers <i>Question Panel</i>	
10.00am-10.30am	Morning Tea	
Genetic Disease Models and Genetics Oversight NZ (joint with Applied Genetic Technologies) Chair Francis Hunter, University of Auckland		
10.30am-10.50am 20 mins	Tracey Dickson (R21) University of Tasmania <i>Getting the Balance Right: Targeting Excitatory Dysfunction in the ALS Cortex</i>	
10.50am –11.10am 20 mins	Imke Tammen (R22) 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	Renee Handley (R23) University of Auckland <i>Brain urea increase is an early pathological event in Huntington's disease</i>	
11.30am-11.50pm 20 mins	Peter Dearden (R24) 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	Kate Neas (R25) 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	Discussion session <i>Sharing/ improving/linking research resources</i>	
12.30pm-1.30pm	Lunch	

Emerging Therapies and keynote Chaired by Prof. Anthony Hannan, University of Melbourne		
1.30pm – 1.50pm 20 mins	Nadia Mitchell (R26) University of Otago <i>Efficacy of gene therapy in a sheep model of CLN5 Batten disease</i>	
1.50-2.10pm 20 mins	Miriam Rodrigues (R27) Auckland District Health Board <i>SMA: Treating the Cause</i>	
2.10-3.00pm 50 mins	Prof. Chris Shaw (R28) <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	All session speakers <i>Question Panel</i>	
3.15pm-3.30pm 15 mins	Meeting close Emma Scotter University of Auckland	
3.30pm-4.30pm 60 mins	MND ROUND TABLE (all stakeholders) <i>Research directions</i>	