# Nanopore Day, Queenstown 2023 Agenda & Speaker Abstracts

**Date: Thursday 31st August 2023**

**Time: 11:00 – 17:30**

**Registration starts at 10:30 am and lunch will be provided.**

**Location: Crowne II room, Crowne Plaza Queenstown Hotel**

**Register here: https://nanoporetech.com/event/NanoporeDayQueenstown**

**Oxford Nanopore Technologies plc**

**nanoporetech.com**

**Find out more on Resource Centre**

# Agenda

| Time | Agenda | Presenter |
|---|---|---|
| 10:30 – 11:00 | Registration | |
| 11:00 – 11:05 | An introduction to Oxford Nanopore | **Warren Bach**<br>Oxford Nanopore Technologies |
| 11:05 – 11:30 | Updates from Oxford Nanopore Technologies | **Mike Yarski**<br>Oxford Nanopore Technologies |
| 11:30 - 11:50 | A novel method to rapidly determine the host genomic context of antimicrobial resistance genes | **Olin Silander**<br>Massey University |
| 11:50 – 12:10 | Untangling tangled cancer genomes | **Cristin Print**<br>University of Auckland |
| 12:10 – 12:30 | High-throughput nanopore sequencing to select new types of Sauvignon Blanc | **Darrell Lizamore**<br>Bragato Research Institute |
| 12:30 – 13:30 | Lunch Mixer | |
| 13:30 – 13:50 | Uncovering the genetic causes of unsolved developmental and epileptic encephalopathies using long-read sequencing | **Denis Nyaga**<br>University of Otago, Wellington |
| 13:50 – 14:10 | Using ONT sequencing to divine the mechanisms for natural activation of transposons in crop species | **Chris Winefield**<br>Lincoln University |
| 14:10 – 14:30 | Application of Oxford Nanopore Sequencing on a New Zealand Parkinson's Disease cohort | **Oscar Graham**<br>University of Otago, Christchurch |
| 14:30 – 15:00 | Afternoon tea | |
| 15:00 – 15:20 | Equitable cancer care: Making the most of the MinION | **Robert Day**<br>University of Otago |
| 15:20 – 15:40 | Oxford Nanopore sequencing for the resolution of de novo copy number variants in patients undergoing preimplantation genetic testing for monogenic disorders (PGT-M) | **Kylie Drake**<br>Canterbury Health Laboratories |
| 15:40 – 16:00 | Generating genetic and epigenetic catalogues for cardio-metabolic risk in the Pasifika Heart Study | **Allamanda Faatoese**<br>Christchurch Heart Institute, University of Otago, Christchurch |
| 16:00 – 16:20 | TBC | **Peter Dearden**<br>University of Otago |
| 16:20 – 16:30 | Closing remarks | **Warren Bach**<br>Oxford Nanopore Technologies |
| 16:30 – 17:30 | Social Mixer (with refreshments) | |

# Olin Silander

## Massey University

### Title

A novel method to rapidly determine the host genomic context of antimicrobial resistance genes

### Abstract

Profiling antimicrobial resistance genes (ARGs) in a sample is an important application of metagenomic sequencing. Equally important is determining the organisms or mobile genetic elements in which ARGs are present. When using Oxford Nanopore sequencing, ARG-containing molecules can be enriched through adaptive sampling or cas9-targeted enrichment. However neither of these methods are efficient. Here we present a PCR-based method to enrich ARGs while maintaining their genomic context. Using this method it is possible to profile the organisms containing ARGs within a sample in less than ten minutes of sequencing.

### Biography

Olin Silander is an Associate Professor at Massey University in Auckland, New Zealand. His research focuses on microbial evolution and genomics. He was part of the team that developed the Midnight protocol, a method for sequencing SARS-CoV-2 genomes using Oxford Nanopore. He has recently published on detecting methylation status in bacteria using Nanopore sequencing and is currently working on the detection of RNA modifications using NAnopore sequencing.

# Cristin Print

## University of Auckland

### Title

Untangling tangled cancer genomes

### Abstract

Precision medicine for cancer patients, and the genomic research that underpins it, has transformed cancer patient care. Until now this field has focused almost exclusively on simple genomic aberrations in tumours that are relatively easy to detect, such as single nucleotide variants, small indels and whole chromosome amplifications. However, we are now trying to expand the precision oncology repertoire into the difficult realm of complex structural variants and their associated copy number aberrations in cancer. This talk will outline our early attempts to untangle structural variants in genetically complex tumours using Oxford Nanopore long-read methodology synergistically with a range of genomic techniques.

### Biography

Cris graduated in Medicine and Surgery from the University of Auckland in 1989 and began research while working as a house surgeon in Dunedin, NZ. A PhD in the University of Auckland led to a four-year postdoctoral fellowship in the Walter and Eliza Hall Institute in Melbourne, Australia before six years in Cambridge University, UK, where he was a Fellow of St Edmunds College and developed a deep interest in genomics and bioinformatics. While there he co-founded a bioinformatics company that became listed on the Tokyo stock exchange in 2007. In 2005 he returned to the University of Auckland where he leads a cross-disciplinary research team of clinicians, cell biologists and data scientists who use genomics, systems biology and bioinformatics to better understand human disease, especially cancer. He leads the Genomics Into Medicine Strategic Research Initiative in Auckland, Chairs the Auckland Regional BioBank Scientific Advisory Board and is a Principle Investigator in the Maurice Wilkins Centre. Until recently he was Acting Chair of the NZ Institute of Environmental Science and Research (ESR) and in the past has served as President of the NZ Society for Oncology and Director of the Bioinformatics Institute at the University of Auckland.

# Darrell Lizamore

## Bragato Research Institute

### Title

High-throughput nanopore sequencing to select new types of Sauvignon Blanc

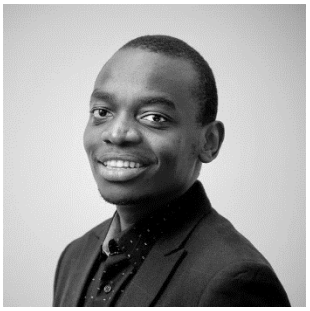
### Abstract

Population-scale whole-genome sequencing has the potential to supercharge functional genomic research in medicine, ecology and agricultural biotechnology. In our work to improve New Zealand's wine grape varieties, transposition events and epigenetic variation underlie much of the novel diversity we are producing. Last year, BRI installed NZ's first P24 sequencer in the hope that high-throughput nanopore sequencing would provide an answer to the daunting task of cataloguing repeat element and epigenetic variation in a very large population. Since then, we have generated a new heterozygous genome reference, tested low-coverage genotyping and compared methylation calling with bisulphite sequencing across cytosine contexts.

### Biography

Dr Darrell Lizamore leads New Zealand's grapevine improvement programme at the Bragato Research Institute. His team works on developing ways to use functional genomic information to improve non-model crops such as wine grapes. A focus of their efforts is understanding epigenetic adaptation to environmental stress and using this knowledge to alter gene expression levels and increase somaclonal diversity. Recently, the team has been trialling the use of high-throughput nanopore sequencing to accelerate crop breeding, and exploring other applications of this technology in New Zealand.

# Denis Nyaga



## University of Otago, Wellington

### Title

Uncovering the genetic causes of unsolved developmental and epileptic encephalopathies using long-read sequencing

### Abstract

Developmental and epileptic encephalopathies (DEEs) are the most severe group of epilepsies affecting 1 in 600 live births. Despite improvements in genetic testing methodologies, >50% lack a precise genetic diagnosis. Thus, better technologies are needed to identify the genetic causes. We sequenced whole genomes of 11 individuals with unsolved DEEs using long-read sequencing (LRS) by Oxford Nanopore Technologies (ONT). We computationally targeted 1,753 epilepsy genes for pathogenic single nucleotide variations (SNVs) and structural variations (SVs). We aimed to demonstrate that LRS enables precise refinement of pathogenic SVs identified by arrays, and may identify causal variants not detected by other sequencing technologies.

### Biography

Dr Nyaga is a postdoctoral fellow at the Department of Paediatrics and Child Health, University of Otago (Wellington, New Zealand). He leads various bioinformatics projects on epilepsy research within the Epilepsy Research Group (ERG). A key focus of his research is to identify the genetic causes of unsolved children with developmental and epileptic encephalopathies (DEEs). Nyaga obtained his PhD in Biomedical Science from the University of Auckland (New Zealand) and his MSc in Translational Oncology from the University of Hull (UK). He also holds a BSc in Molecular and Cellular Biology from Kenyatta University (Kenya).

# Chris Winefield

## Lincoln University

### Title

Using ONT sequencing to divine the mechanisms for natural activation of transposons in crop species

### Abstract

Studying repeat element space in genomes has traditionally been a gargantuan task, complicated by short read whole genome sequencing approaches. ONT sequencing and associated long and ultra long read approaches has been a game changer, not only for resolution of repeat associated issues with de novo genome assembly but importantly to facilitate new and exciting insights into the role of transposons in regulating genome evolution and activity. Our team has been focused on developing a range of do novo assembled genome resources to study the biology of transposons in a wide range of complex diploid and polyploid plant genomes. In this presentation I will summarise the approaches we are currently using and the insights we have develpmed into the role of transposons in generating novel phenotypes in plants and the associated role that transposons plat in regulating epigenetic responses in plant genomes.

### Biography

Chris is currently an Associate Professor at Lincoln University where his team investigates the role of transposons in crop evolution. He is currently an associate investigator at the Australian Research Council, Centre of Excellence in Plant Success and works closely with Prof. Peter Waterhouse at the Queensland Unviersity of Technology investigating the role of transposons in Nicotianana benthamiana responses to climate extremes typical of the natural range of ecotypes of this species. He is also involved in investigation of the role of transposons in Apomixis, specifically in the regulation of parthenogenesis and autonomous endospermy in Mendels Hawkweed. He is also involved with the Bragato Research Institute and the Hapi Research consortia investigating the potential to use activation of transposons as a means of generating novel genetic and phenotypic diversity in grapevine and Hops respectively as well as studying the molecular drivers of sexual dimorphism in Hops. While not sequencing genomes of Nicotiana, Hawkweed, Grapes and Hops he can be found on the many rivers and Lakes ofthe South Island enjoying the scenery and challanges associated with trying to catch trout on the fly.

# Oscar Graham

## University of Otago, Christchurch

### Title

Application of Oxford Nanopore Sequencing on a New Zealand Parkinson's Disease cohort

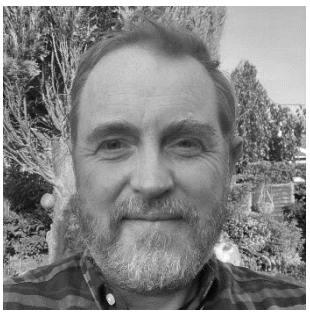
### Abstract

Oxford Nanopore sequencing offers new opportunities for exploring disease causing variants that are difficult to characterise using routine sequencing approaches. Through interrogation of the GBA gene locus (the most common Parkinson's disease associated gene) with long reads we were able to identify several disease-associated variants and establish the haplotype of these variants. The establishment of direct molecular haplotypes lead to discovery of a novel association between common variants inherited together as a haplotype, and Parkinson's disease onset. Additionally, using adaptive sampling we have developed a proof-of-principle method for screening genes associated with neurological disease in patients where the underlying disease aetiology is unknown.

### Biography

I am currently in the final stages of a PhD in biomedical science at the Christchurch campus of the University of Otago. My PhD research revolves around the use of Oxford Nanopore Sequencing for detecting and characterizing variants in Parkinson's disease patients. More broadly I am interested in exploring how novel disease-causing variants can influence clinical and developmental features of neurodegenerative diseases.

# Robert Day



## University of Otago

### Title

Equitable cancer care: Making the most of the MinION

### Abstract

Cancer is New Zealand's single biggest cause of death and each year in Aotearoa, over 25,000 New Zealanders are diagnosed with cancer and >9,000 die from the disease. Survival rates in New Zealand lag many other developed countries, highlighting the need to enable better and more equitable access to treatment. Here I will talk about our efforts to create nanopore-based assays for early cancer detection, monitoring and predicting tumour response to treatment.

### Biography

Rob currently works as a Research Fellow in the Biochemistry Department at the University of Otago. He is a keen advocate for the applied use of genomics-based technologies in the areas of agricultural biology, environmental testing, and medicine. He spends half his time managing the Otago Genomics Facility and is currently developing new methods for sequence-based diagnostics from single cells and circulating tumour DNA in the Centre for Translational Cancer Research.

# Kylie Drake

## Canterbury Health Laboratories

### Title

Oxford Nanopore sequencing for the resolution of de novo copy number variants in patients undergoing preimplantation genetic testing for monogenic disorders (PGT-M)

### Abstract

De novo variants historically excluded patients from accessing PGT-M, as analysis relies on linkage. Recently, whole genome amplification of embryonic cells allowed patients with de novo variants detectable by Sanger sequencing or microarray, to utilise PGT-M. However, de novo 'intermediate' sized variants, single exon to ~10Mb in size, are not easily resolved. Patients with these variants have to date only had the option to undertake prenatal testing of naturally occurring pregnancies. We have utilised Oxford Nanopore long-read sequencing of de novo breakpoints to facilitate PGT-M by coupling direct detection of the breakpoint with single tandem repeat linkage in these patients.

### Biography

Dr Kylie Drake has a PhD from the University of Otago, where she studied the pre-birth origins of childhood leukaemia. Following graduation, she spent 10 years at the Cleveland Clinic in Cleveland, Ohio studying the genetics of thoracic disease. Since 2016 she has been employed as a Scientific Officer at Canterbury Health Laboratories, a busy tertiary referral diagnostic laboratory, where she oversees the exome team, familial testing, and preimplantation genetic testing. Kylie is particularly passionate about the repatriation of diagnostic genetic testing to New Zealand.

# Allamanda Faatoese

### Christchurch Heart Institute, University of Otago, Christchurch

#### Title

Generating genetic and epigenetic catalogues for cardio-metabolic risk in the Pasifika Heart Study

#### Abstract

Cardiovascular disease prevalence is higher in Pacific Peoples than non-Maori, non-Pacific groups in Aotearoa New Zealand. There is a paucity of data that utilises long-read genetic and epigenetic sequencing to understand the inherited risk for Pacific ethnicities. The Pasifika Heart study, a cohort of 200 Pacific adults attempts to generate a genetic and epigenetic catalogue using nanopore sequencing and analysis tools.

#### Biography

Allamanda Faatoese is a Pacific Research Fellow with the Christchurch Heart Institute, University of Otago Christchurch. She completed her PhD studies in 2013 (Department of Medicine, University of Otago) that focussed on biomarkers of heart disease among Aotearoa's indigenous communities in New Zealand. Her postdoctoral fellow studies focussed on building knowledge for Pasifika communities in Aotearoa/New Zealand. She now leads the Pacific Heart Research group that investigates biomarkers (circulating, genetic and epigenetic) of cardiovascular disease and community intervention studies in Pasifika communities.

# Peter Dearden

### University of Otago

#### Title

TBC

#### Abstract

TBC

#### Biography

TBC

# Mike Yarski

### Oxford Nanopore Technologies

#### Title

Updates from Oxford Nanopore Technologies

#### Abstract

Oxford Nanopore Technologies (ONT) has posted roughly 98 product updates within the last 12 months. Kit LSK114 and the R10.4.1 pore flow cells are now in full release. We are looking to streamline and simplify our library kit offerings. With Kit LSK114 ONT sequencing does provide a more complete alternative for large-scale genomics by detecting SNPs and indel calling comparable to established technologies and in addition allows the calling of structural variants and haplotype-specific methylation calls. Overall this provides the most complete picture of the genome on a single platform.

#### Biography

Mike joined ONT in 2019 as a Field Application Scientist to support researchers in Australia, New Zealand and Singapore by providing updates and trainings as the technology develops. Prior to working for Oxford Nanopore, Mike spent over 10 years working for a leading Australia and New Zealand distributor supporting

a range of genomic technologies including Advaita, PacBio, Fluidigm, 10X Genomics, and Affymetrix both as a technical sales specialist as well as an applications specialist. Prior to his commercial roles, Mike completed his PhD at the University of California, Irvine before relocating to Melbourne, Australia where he worked in a few labs as a post-doctorial scientist.