AGTA | Australasian Genomic Technologies Association

2018 ANNUAL CONFERENCE

HANDBOOK

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Tel: 07 3362 0475

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AGTA EXECUTIVE TEAM

Vikki Marshall
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The University of Melbourne

Richard Tothill (Vice President)
The University of Melbourne

Mark Van der Hoek (Treasurer)
South Australian Health & Medical Research Institute

Andreas Schreiber (Vice Treasurer and Membership Secretary)
SA Pathology

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University of Auckland

Rob Day
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The Australian Wine Research Institute

Mark Corbett
The University of Adelaide

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The University of Adelaide

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University of New South Wales

Vikki Marshall
The University of Melbourne

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CONFERENCE • EVENT • ASSOCIATION MANAGEMENT

Welcome from the AGTA President

On behalf of the managing executive of the Australasian Genomic Technologies Association (AGTA), I would like to join conference co-convenors Andreas Schreiber and Mark Van der Hoek in welcoming you to the beautiful seaside suburb of Glenelg, Adelaide for the 18th conference of the association.

AGTA's annual conference, a highlight of the genomics calendar, changes location each year to a new venue within Australia or New Zealand. The location is chosen strategically as a platform for bringing together key national and international experts, students and industry leaders in the field of genomic technologies and applications from around the world.

Each of our annual conferences develops its own unique flavour which comes from the combination of the local genomics research and the invited speakers and students. The 2018 conference promises a wonderful opportunity to network, learn and collaborate in your field of research - we strongly encourage you to enjoy and make the most of the opportunities that this forum provides!

The 2017-2018 year has been productive for AGTA. We welcomed Joseph Powell, head of the Garvan Weizmann Centre for Cellular Genomics and Sam Buckberry, ARC-NHMRC Fellow from the University of WA, to the managing Executive Committee. We farewelled Mark Waltham as President (2015-2017) and Marcel Dinger as a director. A new initiative introduced the support of one AGTA member to attend the AGBT conference in Orlando Florida, by competitive application. In May the executive committee met for its annual planning day to discuss the strategic direction of the Association and we look forward to implementing new initiatives for our members into 2019.

All AGTA members and delegates are encouraged to attend the Annual General Meeting of the Association to be held at this conference. You will to hear about the Associations' achievements, financial position and most importantly, the opportunity to have your say, as we continually strive to improve our offerings to members and to keep abreast of the rapidly changing genomics landscape.

Importantly, we extend a special mention to all our generous and loyal sponsors for their support of our Association's annual conference. These include BGI (Platinum Sponsor), Microsoft (Gold Sponsor), Illumina (Silver Sponsor) and Bronze sponsors Integrated DNA Technologies, Twist BioScience and BD Biosciences. The support of our sponsors is critical to the success of our annual conferences; we would not be able to come together without them. Please ensure that you take the time to visit all trade booths and establish or strengthen networks that will assist you with your research and applications.

We warmly welcome all speakers, and highlight an amazing line up of keynotes from around the world including: Shoji Tsujj (University of Tokyo - neurogenomics); Elizabeth Dinsdale (San Diego State University - marine eco- and evolutionary genomics); Jim Haseloff (University of Cambridge – synthetic biology and reprogramming of plant systems); Kevin White (The University of Chicago – gene networks in disease and evolution); Ludovic Orlando (University of Copenhagen – ancient DNA research); Marlon Stoeckius (New York – technology innovation); Taru Tukiainen (University of Helsinki Genome Center – human genetics and public health, computer and information science).

Look forward to seeing you there!

Vikki Marshall
AGTA President

Welcome from the Conference Convenors

On behalf of the Organising Committee and the Australasian Genomic Technologies Association (AGTA) it is our pleasure to welcome you to Adelaide at the Glenelg seaside for the 18th annual conference of the organization.

The AGTA conference fulfils a unique role in Australasia's yearly genomics conference calendar, and this year is no exception. We will hear about the exciting genomics research going on in laboratories both around the region and beyond. The conference also provides ample opportunity for researchers, core service providers and industry representatives to interact, learn from each other, find out about applications of recent genomic technologies and obtain a first glimpse of what might be just over the horizon.

Adelaide has a vibrant genomics community, working across a wide range of disciplines, and this is reflected in the choice of themes for this conference. Biomedical genomics is well represented in the cancer and neurogenetic sessions, and we will also hear how modern sequencing can help decipher the analysis riddles posed by samples of forensic and ancient DNA origin. We will learn about what the genomic diversity in microbial communities tells us about the ecosystems of the ocean and the gut, and of course will be awed by the daunting challenges of doing genomics in the plant kingdom. As is traditional for AGTA conferences, bioinformatics plays a key role throughout the program. Finally, the New Technologies session, as well as special industry workshops, will give us insights into what is now becoming technologically possible.

We want to give particular mention to our generous sponsors, without whom this conference would not be taking place, and also extend a very special welcome to all our esteemed keynote and invited speakers from around the world.

We hope that during your stay you also have time to take in the beautiful surroundings that the location of the Stamford Grand affords. As well as the beautiful beach and the entertainment areas, the precinct also abounds in historical significance. We are located only metres away from the spot where, 182 years ago, the first European settlers arrived to establish the capital of South Australia (having found Kangaroo Island not to their liking). We trust that you'll agree they chose well!

Welcome to AGTA 2018!

Mark van der Hoek
Conference Convenor

Andreas Schreiber
Conference Convenor

General Information

REGISTRATION DESK

Please direct any questions you may have regarding registration, accommodation or social functions to Leishman Associates staff at this desk.

Registration Desk Opening Times:

Sunday 4 November	11:00 AM – 6:00 PM
Monday 5 November	7:30 AM – 5:30 PM
Tuesday 6 November	7:30 AM – 6:00 PM
Wednesday 7 November	7:30 AM – 5:30 PM

ACCOMMODATION

If you have any queries relating to your accommodation booking first speak to the staff at your hotel or alternatively Leishman Associates staff at the Registration Desk.

Your credit card details were supplied to the hotel you have selected as security for your booking. If you have arrived 24 hours later than your indicated arrival day you may find that you have been charged a fee. You will be responsible for all room and incidental charges on check out and may be asked for an impression of your credit card for security against these charges. This is standard policy in many hotels.

DELEGATE NAME BADGES

All delegates, speakers, sponsors and exhibitors will be provided with a name badge, which must be worn at all times as it is required for access to all the conference sessions and social functions.

With thanks to our Name Badge Sponsor:

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CONFERENCE PROCEEDINGS

Speaker PowerPoints will be available on the AGTA18 website following the conclusion of the conference. Speakers will be requested to sign a release form. This is not compulsory.

CONFERENCE WIFI

Wireless internet will be available throughout the conference venue for the duration of the conference.

TO CONNECT:
Connect to Stamford Wireless
Open your preferred browser
Select that you are a first-time user
Select our conference code

Password: **AGTA2018**

TWITTER

Join the conversation at @agtaGenomics #AGTA18

FAMILY ROOM

As an equity and diversity initiative in support of delegates with parenting or caring responsibilities, a private room will be available at the conference venue for parents with young children, in the Hindmarsh Room. This room is also available for delegates to use as a quiet room if they require a breakout space from the Plenary or Trade. Live audio from the Plenary using headphones, along with presenter PowerPoint presentations, will be available to view from this room.

General Information

EQUITY AND DIVERSITY STATS

The local organizing committee strove to provide a balanced program for AGTA 2018 ensuring gender balance was maintained wherever possible. We were also conscious of the fact that the antipodean version of the tyranny of distance provides extra challenges for our colleagues from WA and New Zealand to attend and contribute. Pertinent statistics include the makeup of the organizing committee itself (5M/5F), the student organizers (2M/1F), session chairs (8M/6F) and national and international invited speakers (11M/8F). Of the 20 submitted abstracts selected for inclusion (10M/10F), 40% were from students or ECRs. 42% of the national invited speakers were from either NZ or WA, and special travel awards were available for attendees from across the Tasman.

DRESS CODES

Dress throughout the day is smart casual or informal business.

EMERGENCY MEDICAL CARE

For any medical emergency please telephone 000. The staff at your hotel will have information if you require contact details for a doctor, dentist or other health professional.

WE HAVE GONE GREEN!

Extra care is being taken this year to ensure that we are not contributing waste and are instead trying to maintain an ecological balance. This is why this year we are not providing delegates with a physical satchel but instead have a Mobile App full of information, prizes and competitions, and material from our sponsors and exhibitors!

There are brown paper bags available for delegates to make collecting sponsor and exhibitor flyers manageable!

EXHIBITOR PASSPORT PRIZE DRAW

This year the exhibitor passport is built in to your Mobile App! The AGTA18 organising committee encourages you to visit each trade exhibitor to scan in and collect interactions, to go into the draw to win some great prizes!

Details of the prizes on offer can be found listed in the Conference App and on display in the prize cabinet. Visit each exhibitors' booth and scan the QR Code to enter into the prize draws on offer. Entries will be collected up until the end of the lunch break on the final day of the conference. Winners will be announced and contacted directly via the Conference App. Prizes need to be collected from the registration desk by the lunch break of the final day.

CONFERENCE APP LEADER BOARD COMPETITION

Delegates will have the ability to engage with the conference in a variety of ways via the Mobile App and by doing this will accrue points/badges. Please refer to the email sent to you prior to the conference that outlines the full functionality of the Mobile App or visit the registration desk if you have any queries. A Leader Board showing the most engaged delegates will be listed in the App, with points being accrued up until the end of the lunch break on the final day of the conference. The top delegate on the Leader Board at the close of the competition will be deemed the winner with an announcement made and a prize awarded in the closing remarks of the conference.

General Information

BARISTA COFFEE

Barista coffee will be available throughout the conference.

Proudly sponsored by 华大基因 BGI

SOCIAL PROGRAM ENTRY

The Welcome Reception is included in the cost of each full conference registration.

The Conference Dinner IS NOT included in any registration type. Social events ARE NOT included in the cost of day registrations or for accompanying partners. Places for day registrants and additional guests for these events may still be available at an additional cost. Bookings can be made at the registration desk subject to availability.

STUDENT FUNCTIONS

All conference students and early career researchers are invited to a casual function on Monday 5 November at Stamford Grand, Moseley Room. If you would like to attend and have not pre-registered, please see Leishman Associates staff at the registration desk. Further information about this event can be found on page 20.

PHOTOGRAPHS, VIDEOS, RECORDING OF SESSIONS

Delegates are not permitted to use any type of camera or recording device at any of the sessions unless written permission has been obtained from the relevant speaker. There will be a professional photographer at the conference and conference dinner. If you do not wish for your photo to be taken, please advise staff at the registration desk.

SPEAKERS AND SPEAKER'S PREPARATION ROOM

All speakers should present themselves to the Speaker's Preparation Room, located in room Colley 2, at least 2 hours before their scheduled presentation time, to upload their presentation.

A technician will be present in the room as follows:

Sunday:	4:00pm - 5:00pm
Monday:	8:00am - 2:00pm
Tuesday:	8:00am - 2:00pm
Wednesday:	8:00am- 1:00pm

Speakers are requested to assemble in their session room 5 minutes before the commencement of their session, to meet with their session chair and to familiarise themselves with the room and the audio visual equipment. For information on the chairperson attending your session, please see the Registration Desk.

ORAL PRESENTATIONS

Please refer to the program for the time allocated for each presentation, as these do vary. The chairperson for your session will give you a 3 minute warning, however you are asked to stick to your time allocation so that the program remains on schedule.

POSTER SESSIONS

Posters will be displayed throughout the foyer and in the Colley Room for the duration of the poster sessions. Posters will only be displayed on the day of their presentation. There will be a poster session on Monday 5 November from 3:30 PM to 4:30 PM and on Tuesday 6 November from 2:20 PM to 3:20 PM

IDT® INTEGRATED DNA TECHNOLOGIES

General Information

SPECIAL DIETS

All catering venues have been advised of any special diet preferences you have indicated on your registration form. Please identify yourself to venue staff as they come to serve you and they will be pleased to provide you with all pre-ordered food. For day catering, there may be a specific area where special food is brought out, please check with catering or conference staff.

SECURITY

The members of the conference organising committee, Leishman Associates and The Stamford Grand accept no liability for personal accident or loss or damage suffered by any participant, accompanying person, invited observer or any other person by whatever means. Nor do we accept liability for any equipment or software brought to the conference by delegates, speakers, sponsors or any other party.

Please protect your personal property. Do not leave laptops, cameras, and other valuable items unsecured. Be conscious of individuals who appear out of place and do not wear a conference name badge. Advise Leishman Associates staff if this does occur.

DISCLAIMER

The 2018 AGTA Conference reserves the right to amend or alter any advertised details relating to dates, program and speakers if necessary, without notice, as a result of circumstances beyond their control. All attempts have been made to keep any changes to an absolute minimum.

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CONFERENCE PROGRAM

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PRE-CONFERENCE WORKSHOPS

1130 - 1530	Species Identification in Metagenomic Data Sets: From the Clinic to the Ocean **Elizabeth Dinsdale**, San Diego State University Metagenomic assembly and identifying environmentally important proteins **Ludovic Orlando**, University of Copenhagen Tentative title: A pipeline for efficient species ID **Rob Edwards** Metagenomics in the cloud **Laura Weyrich**, University of Adelaide Contaminating DNA problems in Metagenomic Research **Room:** Moseley 1-3
1130 - 1530	Galaxy Australia – The Free Genetics and Genomics Analysis Platform **Gareth Price** **Room:** Main Plenary Hall
1100 – 1800	Conference Registration Open

OPENING ORATION

Chair: Vikki Marshall

1700 - 1800	**Guest Speaker - Kevin White**
1800 - 2000	**Welcome Reception & Exhibition** Stamford Grand

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Today's Barista Cart is Sponsored by:	华大基因 BGI
0730 - 1730	Registration Desk Open
0800 - 1800	Exhibition Open
0800 - 0830	Industry Session **Jay Patel** Eliminating the bioinformatics bottleneck: Rapid and accurate analysis at genomic scale illumina®
0845 - 0900	**Official Welcome and Conference Opening**
SESSION 1: Ancient DNA & Forensics Chairs: Laura Weyrich & Helen Speirs	
0900 - 0945	**Ludovic Orlando** Tracking Six Millenia of Horse Selection, Admixture and Management with Complete Genome Time-Series
0945 - 1010	**SallyAnn Harbison** Challenges and opportunities in transforming Forensic DNA genotyping to massively parallel sequencing technology
1010 – 1035	**Jeremy Austin** Using hybridisation enrichment and next generation sequencing for identification of highly degraded human remains.
1035 - 1105	**Morning Refreshments & Exhibition**
1105 – 1130	**Michael Knapp** New technologies for an old problem - the role of ancient DNA and functional genomics in bird conservation
1130 - 1150	**Vanessa Hayes** Analysis of 1025 L0-mitogenomes suggest a geographical divide shaped early human dispersals within Southern Africa
1150 - 1200	Lightning Poster Session 1
1200 - 1300	**Lunch & Exhibition**
SESSION 2: Neurogenetics Chairs: Mark Corbett & Sam Buckberry	
1300 - 1345	**Shoji Tsuji** Non-coding repeat expansions consisting of same repeat motifs in three genes cause benign adult familial myoclonic epilepsy.
1345 - 1410	**Naomi Wray** Genetics of Common Disease: Progress and Next Steps

1410 - 1430	**Peter Hickey** Genome-wide analysis of DNA methylation in samples from the Genotype-Tissue Expression (GTEx) project	●
1430 - 1450	**Akira Gokoolparsadh** The Landscape of Circular RNA Expression in the Human Brain	● ●
1450 - 1510	**Anushree Balachandran** Uncovering novel aspects of human neural crest biology using multiomic approaches	●
1510 - 1530	**Natalie Twine** Big Data Technologies Enables Distant Relationship and Disease Variant Discovery in ALS	● ●
1530 - 1630	**Poster Session 1 & Afternoon Refreshments & Exhibition**	IDT INTEGRATED DNA TECHNOLOGIES
SESSION 3: New Technologies Part 1 Chairs: Mark Van der Hoek & Vikki Marshall		
1630 – 1700	**Radoje Drmanac** DNA Nanoballs and Single-Tube LFR: Affordable "Perfect" Genome Sequencing	华大基因 BGI ●
1700 - 1720	**Quentin Gouil** Reconstructing haplotyped methylomes with long-read sequencing for fine dissection of imprinting in mice	● ●
1720 - 1740	**Martin Smith** Massively multiplexed targeted long-read single cell sequencing for deep phenotyping of tumour and immune cell repertoire	●
1740 - 1800	**Eva Chan** Cool things that can be done with Optical Mapping	●
1800 - 2000	**Student Night – Students Only**	AGTA Australasian Genomic Technologies Association
1800 - 2100	**VIP Function – Invitation Only**	AGTA Australasian Genomic Technologies Association

SPEAKER KEY:

● Invited Abstract ● Student ● Early Career Researcher ● National Invited Speaker ● Keynote

Today's Barista Cart is Sponsored by:	华大基因 BGI
0730 - 1800	Registration Desk Open
0800 - 1800	Exhibition Open
0800 - 0830	Industry Session **Felipe Ayora - Cloud Solution Architect** How researchers are using the Microsoft cloud to accelerate genomics analyses Microsoft
0850 - 0900	**Welcome to Day Two**
SESSION 4: New Technologies Part 2 Chairs: Mark Van der Hoek & Vikki Marshall	
0900 - 0945	**Marlon Stoeckius** CITE-seq: Highly multiplexed simultaneous measurement of epitopes and transcriptomes in single cells BD
0945 - 1010	**Ryan Lister** Dissecting plant development at single cell resolution
1010 - 1040	**Morning Refreshments & Exhibition**
SESSION 5: Cancer Chairs: Deb White & Vanessa Hayes	
1040 - 1125	**Kevin White** From Petabytes to Molecular Insights: Building a System for Precision Medicine
1125 – 1145	**Sujanna Mondal** The super enhancer-driven long noncoding RNA lncNB promotes neuroblastoma tumorigenesis
1145 - 1205	**Katherine Pillman** MicroRNAs regulate large-scale alternative splicing in cancer by controlling the RNA-binding protein Quaking
1205 – 1215	Lightning Poster Session 2
1215 - 1315	**Lunch & Exhibition**
1315 – 1340	**Nic Waddell** Whole Genome Sequencing of Tumour and Normal Samples to Identify Cancer Aetiology and Therapeutic Opportunity
1340 - 1400	**Sunny Wu** Landscape of the breast cancer tumour microenvironment using single-cell RNA sequencing

1400 - 1420	**Richard Tothill** Going nuclear: Using single-nuclei RNA-sequencing for analysis of rare tumours
1420 - 1520	**Poster Session 2 & Afternoon Refreshments & Exhibition & Screening of Melbourne Cup**
SESSION 6: Bioinformatics Chairs: Andreas Schreiber & Kat Pillman	
1520 - 1605	**Taru Tukiainen** Genomic approaches to decipher the extent and impacts of X chromosome inactivation
1605 - 1630	**Nicole Cloonan** Form versus Function: Understanding the beauty in genomic noise
1630 - 1650	**Harriet Dashnow** STRetch: detecting and discovering pathogenic short tandem repeat expansions
1650- 1710	**Hyun Jae Lee** Integrated pathogen load and dual transcriptome analysis of systemic host-pathogen interactions in severe malaria
1710 - 1730	**Hardip Patel** National Centre for Indigenous Genomics
1730 - 1815	**AGTA Annual General Meeting**
1830 - 2330	**Conference Dinner**

SPEAKER KEY:

Invited Abstract · Student · Early Career Researcher · National Invited Speaker · Keynote

Today's Barista Cart is Sponsored by:	华大基因 BGI
0730 - 1730	Registration Desk Open
0800 - 1800	Exhibition Open
0855 - 0900	**Welcome to Day Three**
SESSION 7: Plants Chairs: Matthew Gilliham & Juan Carlos Sanchez Ferrero	
0900 - 0945	**Jim Haseloff** Simple model plant systems for whole organism engineering Microsoft
0945 - 1010	**Jacqueline Batley** Investigating the Role of Structural Variation in Brassica Disease Resistance
1010 - 1030	**Agnieszka Golicz** Roles of long non-coding RNAs in Plant Sexual Reproduction
1030 - 1100	**Morning Refreshments & Exhibition**
1100 - 1125	**Rachel Burton** Genome Sequencing, Assembly and Annotation of the Industrially Useful Plant Plantago ovata
1125 - 1145	**Michael Roach** Population sequencing reveals clonal diversity and ancestral inbreeding in the grapevine cultivar Chardonnay
1145 - 1205	**Yayu Wang** Functional Structure and Association of Root Microbiome with Foxtail Millet Yield Unravelled by Ultra-Deep Metagenomic Sequencing
1205 - 1305	**Lunch & Exhibition**
SESSION 8: Metagenomics Chairs: Renee Smith & Anthony Borneman	
1305 – 1350	**Elizabeth Dinsdale** Mosaic of microbes: Investigating the involvement of marine microbiomes in the life and death of kelp forests
1350 – 1415	**James Paterson** A TCE contaminated aquifer reveals a Piggyback-the-Persistent viral strategy

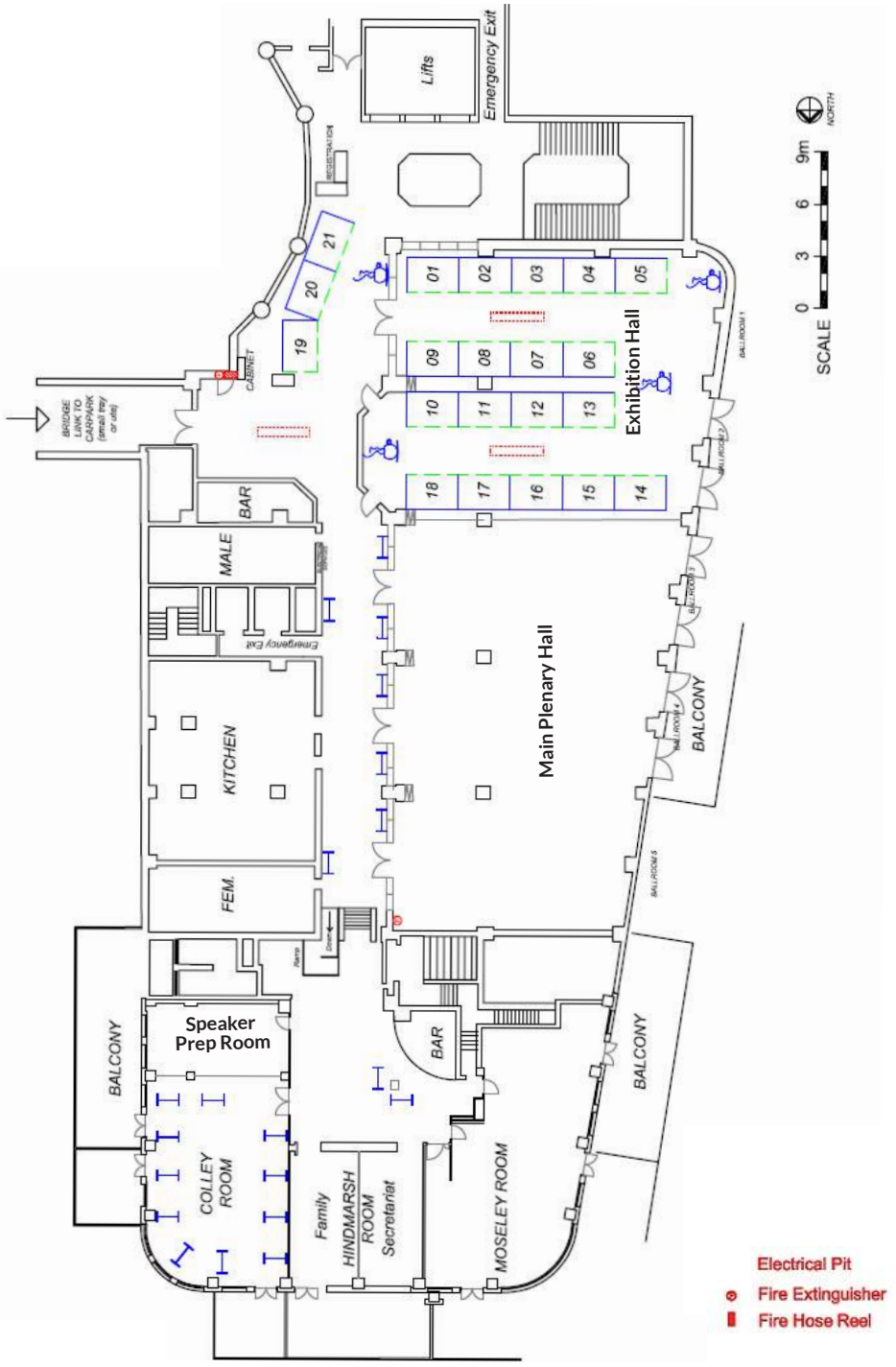
1415 – 1440	**Michael McDonald** Tracking evolution in experimental populations of microbes using time-resolved whole genome sequencing	●
1440 – 1500	**Alicia Byrne** A genomic autopsy of perinatal death	● ●
1500-1520	**Samuel Forster** Genome based metagenomic analysis using an extensive human gastrointestinal culture collection	●
1520 – 1545	**Gene Tyson** Expanding our view of microbial communities in thawing permafrost	●
1545 – 1615	**Awarding of Prizes, 2019 Conference Launch and Conference Close**	

SPEAKER KEY:

● Invited Abstract ● Student ● Early Career Researcher ● National Invited Speaker ● Keynote

Image ©SATC

Floor Plan



EXHIBITORS

Agilent Technologies	2	Illumina Australia	13	New England Biolabs	20
AGRF	8	Integrated DNA Technologies	7	Novogene	12
BD	10	Macrogen Oceania	16	Oxford Nanopore Technologies	22
BGI Australia	4-5	Merck	19	PerkinElmer	1
Bio-Strategy	18	Microsoft	14	Ramaciotti Centre For Genomics	15
Decode Science	6	Millennium Science	3	Trendbio	11
Genewiz	17	Nanostring Technologies	21	Twist Bioscience	9

Conference Social Program

Welcome Reception & Trade Exhibition

The Welcome Reception is included in a full registration only. Additional tickets can be purchased at $75.00 per person.

Date:	Sunday 4 November 2018
Venue:	Ballroom 1 & 2, The Stamford Grand
Time:	6:00 PM – 8:00 PM
Dress:	Smart Casual

Join us for the official Welcome Reception for the 2018 AGTA Conference. Enjoy networking with old and new acquaintances, meeting our sponsors and trade exhibitors, whilst enjoying drinks and canapés.

Student/ECR Function

Students and early career researchers are invited to attend the student night at The Stamford Grand, Moseley Room. Platter style food and a limited beverage service will be provided, don't miss your get to know your peers. Bookings are essential.

Date:	Monday 5 November 2018
Venue:	Stamford Grand, Moseley Room
Time:	6:00 PM – 8:00 PM

VIP Function

Date:	Monday 5 November 2018
Venue:	Sammy's on the Marina
Time:	6:00 PM – 9:00 PM
Cost:	N/A Invitation Only

Conference Dinner

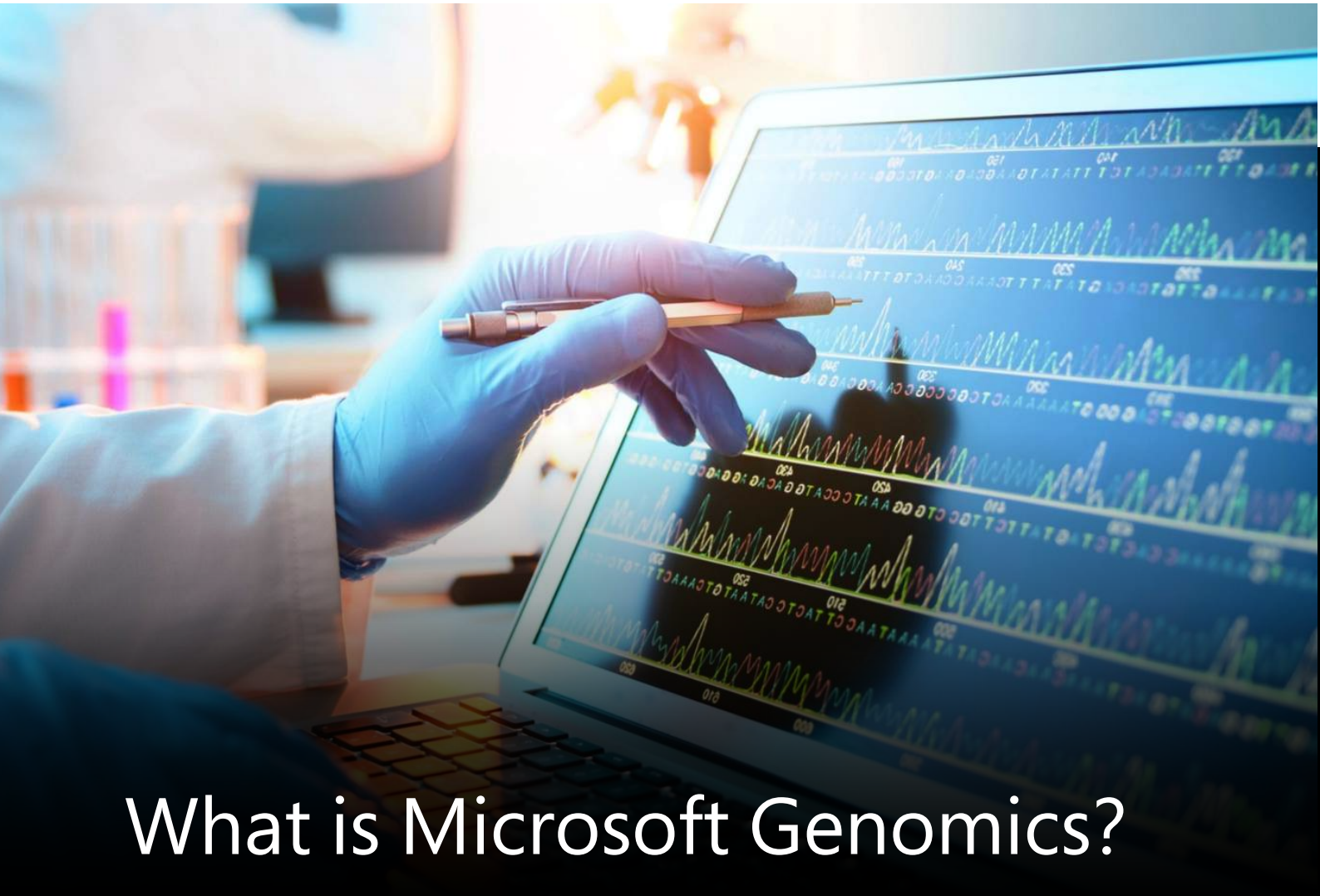
We have worked hard to produce a social event that showcases Adelaide. The social highlight, the conference dinner at Adelaide Oval shouldn't be missed. Transport to the dinner will be provided from The Stamford Grand Adelaide to Adelaide Oval departing at 6.30 PM. Return Transport will also be provided following the conclusion of the dinner.

Tickets to the dinner are essential!

Date:	Tuesday 6 November 2018
Venue:	Adelaide Oval
Time:	6:30 PM – 11:30 PM
Dress:	Smart Casual
Registration:	Tickets to the Conference Dinner could be purchased through the online registration form. Tickets can still be purchased, subject to availability.
Cost:	$165 per ticket. The Conference Dinner is not included in any registration type.
Seating:	The arrangement for this event is free seating. If you have a dietary requirement, please see venue staff.

With Thanks to our Dinner Sponsor

Microsoft



What is Microsoft Genomics?

Microsoft Genomics offers a cloud implementation of the Burrows-Wheeler Aligner (BWA) and the Genome Analysis Toolkit (GATK) for secondary analysis. The service is ISO-certified and compliant with HIPAA regulations, and offers price predictability for your genome sequencing needs. Learn how to use the Microsoft Genomics service and integrate with our API by reading our quickstarts, tutorials, and documentation.

Support your most demanding sequencing needs

Instead of managing your own datacenters, take advantage of the scale and experience of Microsoft in running exabyte-scale workloads. Our cloud implentation of the BWA-GATK is highly concordant with the Broad Institute's best practices pipeline.

Keep your business running

Microsoft Genomics offers a 99.99% availability service level agreement (SLA) for receiving workflow requests.

Secure your data

The Microsoft Genomics service is ISO 27001, ISO 27018, and ISO 9001 certified and compliant with HIPAA regulations.

For more information on workflow SLAs and Azure's platform security visit **azure.microsoft.com**

Microsoft

Pre-Conference Workshops

Sunday 4 November 2018 | The Stamford Grand

The workshops are optional, tickets can be purchased for $40.00 per delegate and $20.00 per student delegate, ticket price includes lunch and afternoon refreshments. Bookings are essential.

PRE-CONFERENCE WORKSHOP 1

Species Identification in Metagenomic Data Sets: From the Clinic to the Ocean

Moseley Room 1-3
11:30 AM – 3:30 PM

Elizabeth Dinsdale, San Diego State University
Metagenomic assembly and identifying environmentally important proteins

Ludovic Orlando, University of Copenhagen
Tentative title: A pipeline for efficient species ID

Rob Edwards
Metagenomics in the cloud

Laura Weyrich, University of Adelaide
Contaminating DNA problems in Metagenomic Research

Speaker Bio

Dr Weyrich received a PhD in Microbiology and Bioethics from Penn State, studying how respiratory infections alter the microbiome. In 2012, she moved to the University of Adelaide and established a research team at The Australian Centre for Ancient DNA that uses calcified dental plaque to reconstruct ancient human oral microbiomes. Her team was the first to reconstruct the microbiome of an extinct species - Neandertals - and has reassembled the oldest microbial genome to date at 48,000 years old. Her team's research has been featured by the BBC, NPR, Science, Nature, New Scientist, NY Times, Smithsonian Magazine, National Geographic, and many others, and has even had a Buzz Feed quiz written about it. Her team is now reconstructing the evolutionary history of the human oral microbiome on six continents, obtaining insight into how the lifestyles and diets of our ancestors impact our health today.

PRE-CONFERENCE WORKSHOP 2

Galaxy Australia – The Free Genetics and Genomics Analysis Platform

Main Plenary Ballroom 3-5
11:30 AM – 3:30 PM

Workshop Speaker
Gareth Price

Speaker Bio

Dr Gareth Price has been a bioinformatician and genomics scientists for over 15 years, working at the Peter MacCallum Cancer Institute, Murdoch Childrens Research Institute, Mater Misericordiae Research Institute, Mater Health Services and Queensland Facility of Advanced Bioinformatics. He has involved in experimental design, assay performance, data QC, data analysis and data interpretation from early printed microarrays, to cartridge based GeneChips through to the latest Next Gen Sequencing platforms. Gareth's publications cover a range of disease models and genetic investigations in a variety of model organisms from microorganisms, fruit flies, mice to humans. Working across a diverse range of biological studies has meant Gareth has acquired a deep understanding of a broad range of bioinformatics skills and applications. This he has applied for the last year and a half at QFAB where Gareth leads the Computational Biology team, helping researchers by performing bioinformatics analyses on their data and providing insight by linking his knowledge of the most appropriate tools or connections between data sources to maximise data value.

ABSTRACTS & BIOGRAPHIES

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SESSION 1:
Ancient DNA & Forensics

Chairs: Laura Weyrich & Helen Speirs

9:00 AM - 9:45 AM

Tracking Six Millenia of Horse Selection, Admixture and Management with Complete Genome Time-Series

Keynote Speaker

Professor Dr Ludovic Orlando
University of Copenhagen

BIOGRAPHY:

Ludovic was born with the first DNA sequencing instruments. He defended his PhD 20 years after the first ancient DNA molecule was sequenced. He was appointed at the age of 27 as an Associate Professor at the Ecole Normale Superieure of Lyon, one of the top-5 French universities. He started his own research group in 2010 at the Centre for GeoGenetics, Univ. of Copenhagen, Denmark, where he has been appointed as a full Professor in Molecular Archaeology since 2016. He became the same year a CNRS research director at University of Toulouse, where he developed AGES, a new multidisciplinary research institute integrating Archaeology, Genomics, Evolutionary biology and Sociology. Research at AGES is aimed to reveal how much such major pasts transitions have impacted our evolutionary trajectory, shaped our biology as well as our health, and how our activities have ultimately circled back to our environment and the communities of species we interact with. Amongst his early career achievements feature (1) the sequencing of the oldest genome, (2) the characterization of the first ancient epigenome, and (3) the reconstruction of the genomic history of horse domestication.

ABSTRACT:

Ludovic ORLANDO[1,2]

[1] Laboratoire d'Anthropobiologie Moléculaire et d'Imagerie de Synthèse, CNRS UMR 5288, Université de Toulouse, Université Paul Sabatier, 31000 Toulouse, France
[2] Centre for GeoGenetics, Natural History Museum of Denmark, University of Copenhagen, 1350K Copenhagen, Denmark

The domestication of the Horse and its impact on warfare, transportation and agriculture, have revolutionized human history. Even though most modern breeds have been engendered within the last couple of centuries, humans have managed horse livestock for over five millenia. Recent selective and management strategies have tremendously impacted the genetic structure of horse populations. As a result, modern patterns of genetic diversity can only partly help reconstruct the horse domestication process prior to the modern era. Recent research in our laboratory, carried out in the framework of the ERC PEGASUS programme, has endeavored to sequence complete horse genomes from across their whole temporal and geographical domestication range in order to identify how the many past human cultures progressively forged the horse genome by means of selection, drift and admixture. This work revealed two different dynamics at play within early and late domestication stages, involving the selection for different functional pathways, different management strategies for the genetic resource available, including stallion diversity, and a recent increase in the genomic deleterious load. Our new genome dataset now allows us to document such changes at unprecedented scales and reveals unexpected features of the whole population dynamic underlying horse domestication, in relation to major human expansion across the steppes.

KEY WORDS

Ancient DNA, Domestication, Horse

9:45 AM - 10:10 AM

Challenges and opportunities in transforming Forensic DNA genotyping to massively parallel sequencing technology

Invited Speaker

Dr SallyAnn Harbison

Institute of Environmental Science and Research

BIOGRAPHY:

Dr SallyAnn Harbison is a Senior Science Leader and the DNA Technical Leader of the Forensic Biology Group of ESR at Mt Albert, actively engaged in forensic casework. Before joining ESR in 1988 SallyAnn completed her PhD at the University of Liverpool in the UK in plant virology and was a postdoctoral research fellow at University of Auckland. From 1988, SallyAnn undertook crime scene, evidence examination and DNA profiling, before specializing in DNA analysis in 1997. She has been an Honorary Lecturer on the Post-graduate Forensic Science Course at the University of Auckland since 1996 and supervises many research students. SallyAnn is currently the Chair of the Biology Special Advisory Group to ANZFEC (Australia and New Zealand Forensic Executive Committee). Her current areas of research interest include genomic analysis of short tandem repeats and SNPs of forensic relevance leading to a transformation to new generation sequencing approaches for casework and the recovery and identification of biological evidence using RNA techniques.

ABSTRACT:

SallyAnn Harbison, Ryan England, Kate Stevenson, Jayshree Patel, Lisa Melia and Alex Liu.

Institute of Environmental Science and Research

Worldwide, forensic genotyping still relies on the analysis of short tandem repeats and capillary electrophoresis for forensic genotyping. However, a small number but growing number of laboratories including our own have embraced next generation sequencing, recognising the potential for increased investigative intelligence information and greater discrimination. This transformation is however not without substantial challenges. In this presentation I will firstly outline the current landscape including statistical models used for data interpretation, the use of DNA profile databases and the quality assurance measures required. I will then describe the challenges faced by the forensic community in moving towards a sequence based approach including nomenclature, bioinformatics, transformation of DNA Databases and method validation to ensure reliability and reproducibility. I will highlight key aspects from our own work in validating and implementing the ForenSeq DNA signature prep kit from Verogen on the MiSeq FGx. I will conclude by commenting on the opportunities arising from expanding the range of genomic based tests and new sequencing technologies.

KEY WORDS

Forensic genotyping, bioinformatics, validation

10:10 AM - 10:35 AM

Using hybridisation enrichment and next generation sequencing for identification of highly degraded human remains.

Invited Speaker

Associate Professor Jeremy Austin
University of Adelaide, Adelaide, Australia

BIOGRAPHY:

Jeremy is an evolutionary biologist working in the fields of ancient DNA, molecular ecology and forensic biology. He received his PhD in from the University of Tasmania in 1995 and has held postdoctoral research positions at the Natural History Museum, University of Queensland, Museum Victoria and the University of Adelaide. In 2011 he was awarded a prestigious Australian Research Council Future Fellowship and is now a Senior Lecturer in the School of Biological Sciences, and Deputy Director of ACAD, at the University of Adelaide. Jeremy's research uses ancient and modern DNA techniques to develop and apply new methods for identification of highly degraded human remains. He has provided DNA testing and expert evidence to Australian police and the Australian Defence Force.

ABSTRACT:

Jeremy Austin

University of Adelaide

DNA based human identification is both critical and central to criminal and coronial investigations, disaster victim and missing persons identification, repatriation of war dead and counter-terrorism operations. Persistent technological and conceptual issues associated with the application of standard procedures to trace and degraded DNA, or when profiles have no match in DNA databases, limit their application to real world human identification problems. I will discuss the development and application of a novel targeted sequence capture and high throughput DNA sequencing approach to simultaneously type thousands of informative identity, ancestry and phenotype markers in a single assay to facilitate identification of unknown suspects and missing persons. This will deliver new capacity and capability in specialist DNA-based identification of trace and highly degraded DNA. This new approach will result in improved and more reliable identification from trace sources and decomposed human remains and will reduce costs and delays in the identification of unknown samples, improving outcomes in criminal and coronial investigations.

11:05 AM - 11:30 AM

New technologies for an old problem - the role of ancient DNA and functional genomics in bird conservation

Invited Speaker

Dr Michael Knapp
University of Otago

BIOGRAPHY:

Dr Michael Knapp is a Rutherford Discovery Fellow and head of the Molecular Ecology and Palaeogenomics lab at the University of Otago. He was awarded his PhD from Massey University, New Zealand and has conducted postdoctoral fellowships at the Max Planck Institute for Evolutionary Anthropology in Leipzig, Germany and at the University of Otago. He has also led a palaeogenetics and molecular ecology research group at Bangor University, UK. Michael has received a number of research awards, including a Royal Society of New Zealand Rutherford Discovery Fellowship and an Illumina Emerging Researcher Award. Michael's research group focuses on how species interact with their environment both on the molecular and on the population level and on how human activity shapes biodiversity.

ABSTRACT:

Michael Knapp[1]

[1] Department of Anatomy, University of Otago, Dunedin, NZ

Conservation management relies on high quality genetic data, but the availability of such data has long been a limiting factor. Recent advances in DNA sequencing technology have the potential to overcome these limitations. Improvements in ancient DNA technologies for example allow us to study extinct species on a population level and over tens of thousands of years to reconstruct potential causes for extinction. Such information can provide valuable insights into potential threads for endangered species today. Furthermore, the ongoing reduction in costs of genome sequencing now allows us to replace neutral genetic markers as proxies for functional variation in a threatened species with actual information on functional genomic diversity. Such data can directly be used in conservation management, for example when making translocation decisions.
I will present ongoing research into extinct and endangered bird species and discuss implications of these studies for the future of conservation genetic research.

KEY WORDS:

Palaeogenomics, extinction, bird conservation

11:30 AM - 11:50 AM

Analysis of 1025 L0-mitogenomes suggest a geographical divide shaped early human dispersals within Southern Africa

Invited Abstract

Prof Vanessa Hayes
Garvan Institute of Medical Research

BIOGRAPHY:

Vanessa Hayes is the Petre Chair of Prostate Cancer Research at the University of Sydney and is Head and Professor of the Laboratory for Human Comparative and Prostate Cancer Genomics at the Garvan Institute of Medical Research. Her research interest is in using the power of genomics to uncover the story of modern history from our early origins to significant adaptations, while uncovering genomic signatures and more recently the microbial landscape associated with the onset, evolution and ultimately treatment for prostate cancer.

ABSTRACT:

Eva K.F. Chan[1], M.S. Riana Bornman[2], Vanessa M. Hayes[1,2,3]

[1] Laboratory for Human Comparative and Prostate Cancer Genomics, Genomics and Epigenetics Division, Garvan Institute of Medical Research, Darlinghurst, NSW 2010, Australia
[2] School of Health Systems and Public Health, University of Pretoria, Pretoria, South Africa
[3] Central Clinical School, University of Sydney, Camperdown, NSW, Australia

Southern Africa is home to contemporary populations, including the click-speaking hunter-gatherer KhoeSan, representing not only the greatest within and between genome diversity, but also the earliest branches of the human phylogenetic tree. As such, southern Africa is a contender for the origin of anatomically modern humans. While whole genome data is largely lacking for the region, mitogenomes have provided a valuable resource to establish southern African prehistory and human dispersals.

Globally rare, the L0 macro-haplogroup represents the deepest rooting mitochondrial branches, having emerged well over 150 thousand years ago (kya). Here we sequence 197 novel and rarely represented southern African L0 mitogenomes, the largest of its kind to date. Combining our data with that published for a total of 1,025 complete L0 mitogenomes, we redefine early human maternal timelines and dispersals. We show that southern Africa was a hub of human activity since the emergence of L0d (the most ancient and common to KhoeSan haplogroup) almost 200 kya, followed roughly 10 thousand years later by the emergence of L0k (KhoeSan-specific haplogroup). Further divergence of the relatively rare L0f occurred 160 kya from L0a (haplogroup common to south-east and central Africa) and the new and extremely rare KhoeSan-specific sister-haplogroup L0g. We observe for the first time a geographic divide, we term the 'Zambezi Divide', that has segregated human dispersals and evolution in southern Africa. We have identified and provided timelines for both novel and reclassified L0d, L0f and L0g lineages that represent independent human dispersals of relevance to the 'Zambezi Divide'. Of significance, we observe the emergence of 12 southern African specific maternal lineages at the time modern humans were migrating out-of-Africa in the north-east of the continent.

KEY WORDS

Mitogenomes, early human origins and divergence, human timelines, southern Africa, KhoeSan

SESSION 2: Neurogenetics

Chairs: Mark Corbett & Sam Buckberry

1:00 PM - 1:45 PM

Non-coding repeat expansions consisting of same repeat motifs in three genes cause benign adult familial myoclonic epilepsy.

Keynote Speaker

Professor Shoji Tsuji

The University of Tokyo Hospital

BIOGRAPHY:

Dr. Tsuji received his MD and PhD from The University of Tokyo. He served his residency in Neurology at Jichi Medical School. In 1984, he moved to NIH and elucidated molecular basis of Gaucher disease. In 1987 he moved to Niigata University, Japan and was appointed as Professor of Neurology and Director at Brain Research Institute, Niigata University. In 2002, he moved to The University of Tokyo as Professor and Chair of Department of Neurology in 2002. His research focus is elucidation of molecular basis of hereditary as well as sporadic neurological diseases based on comprehensive genome analysis. In 2013, his team discovered that COQ2 gene is a susceptibility gene for multiple system atrophy (MSA), which suggested the efficacy of supplementation of a high dose CoQ10 for MSA. He has started a phase 2 clinical trial for MSA this year. Very recently his team discovered noncoding repeat expansions in benign adult familial myoclonus epilepsy (BAFME).

1981	*Associate in Neurology, Jichi Medical School.*
1984	*Visiting Fellow, National Institutes of Health*
1991	*Professor and Chair, Department of Neurology, Brain research Institute, Niigata University*

2001 Director, Brain Research Institute, Niigata University
2002 Professor and Chair, Department of Neurology, The University of Tokyo
2018 Professor, International University of Health and Welfare
2018 Director, Institute of Medical Genomics, International University of Health and Welfare
2018 Project Professor, Department of Molecular Neurology, The University of Tokyo

ABSTRACT:

Shoji Tsuji

Department of Molecular Neurology, Graduate School of Medicine, The University of Tokyo and International University of Health and Welfare

Unstable tandem repeat expansions have been shown to be involved in a wide variety of neurological diseases. Given a rapidly increasing number of diseases belonging to this group, it is expected that many more diseases await identification of causative genes. We have recently identified non-coding repeat expansions in benign adult familial myoclonic epilepsy (BAFME), an autosomal dominant disorder characterized by infrequent epilepsy and myoclonic tremor. The clue for identification of non-coding repeat expansion was obtained by an observation of a TTTTA pentanucleotide repeat located in intron 4 of SAMD12, which showed an apparently inconsistent transmission pattern in a family. An intensive search of the whole genome sequence data of affected individuals revealed TTTCA and TTTTA repeat expansions in intron 4 of SAMD12, which were found exclusively in the patients in the 50 families. Single-molecule, real-time sequencing of BAC clones and nanopore sequencing of genomic DNAs revealed expanded repeat configurations in SAMD12. Intriguingly, in the two families with the clinical diagnosis of BAFME where no repeat expansions in SAMD12 were observed, similar TTTCA and TTTTA repeat expansions were further identified in introns of TNRC6A and RAPGEF2, indicating that expansions of the same repeat motifs are involved in the pathogenesis of BAFME regardless of the genes where the expanded repeats are located.

Based on these findings, we have established the new strategy for identification of expanded repeat expansions in the short reads obtained by massively parallel sequencers. We are currently applying this strategy to identify non-coding repeat expansions in neurological diseases.

1:45 PM - 2:10 PM

Genetics of Common Disease: Progress and Next Steps

Invited Speaker

Naomi Wray
The University of Queensland

BIOGRAPHY:

Professor Naomi Wray holds joint appointments at the Institute for Molecular Bioscience (IMB) and the Queensland Brain Institute (QBI) within the University of Queensland. She is a National Health and Medical Research Council Principal Research Fellow and a Fellow of the Australian Academy of Science. Her research focusses on development of quantitative genetics and genomics methodology with application to psychiatric and neurological disorders. She plays a key role in the International Psychiatric Genomics Consortium and co-leads the IceBucket Challenge funded sporadic ALS Australia systems genomics consortium (SALSA)

Naomi Wray, Prof Peter Visscher and Prof Jian Yang together comprise the Executive Team of the Program in Complex Trait Genomics (PCTG) funded as an NHMRC Program Grant 2017-2021. The PCTG comprises a critical mass of more than 30 post-doctoral researchers plus research assistants and students, all supported by external grant funding. PCTG is structured into five research themes: Statistical Genomics, Systems Genomics, Psychiatric Genomics, MND Genomics and Genomics of Cognitive Ageing.

ABSTRACT:

Naomi R Wray[1,2]

1 Institute for Molecular Bioscience, The University of Queensland, Brisbane, Australia
2 Queensland Brain Institute, The University of Queensland, Brisbane, Australia

The last decade of genome-wide association studies (GWAS) has generated a remarkable range of discoveries in population and complex trait genetics, and biology of diseases. Integration of GWAS results of common disease with GWAS results of molecular traits, including those measured at single cell level, facilitates functional interpretation in silico. In the next ten years, GWAS data are likely to be available of millions of samples with array data imputed to a large fully sequenced reference panel and 100,000s samples with whole genome sequence (WGS) data. A key end-goal is the translation towards new therapeutics or prevention strategies. Treatments or interventions tailored to individuals in so-called Precision Medicine is viewed as the future of clinical practice. A key question is how will we stratify patients for these personalised approaches. Based on the new understanding about the genetic architecture of common disease, I will consider the research strategies that are needed to better enable patient stratification.

KEY WORDS

Common Disease, Genome-wide Association Study, Methylation-wide Association Study, Selection, Stratification, Precision Medicine

2:10 PM - 2:30 PM

Genome-wide analysis of DNA methylation in samples from the Genotype-Tissue Expression (GTEx) project

Invited Abstract

Dr Peter Hickey
Johns Hopkins University

BIOGRAPHY:

Peter Hickey is currently a postdoctoral research fellow in the Department of Biostatistics at Johns Hopkins University working in the research group of Kasper Hansen where he develops statistical methods and software to summarise and understand genomics data.

He enjoys making sense of data and making it easier for others to do the same.

He is excited to be moving back to Australia to take up a position as Senior Research Officer in the single-cell genomics group at the Walter and Eliza Hall Institute of Medical Research.

ABSTRACT:

Peter F. Hickey[1,*], Lindsay F. Rizzardi[2,3,*], Rakel Tryggvadottir[2], Colin Callahan2, Adrian Idrizi[2], Kimberly Stephens[2,4], Sean Taverna[2,4], eGTEx Project, Kasper D. Hansen[1,2,5,+], Andrew P. Feinberg[2,3,6,+]

1 Department of Biostatistics, Bloomberg School of Public Health, Johns Hopkins University
2 Center for Epigenetics, Johns Hopkins University School of Medicine
3 Department of Medicine, Johns Hopkins University School of Medicine
4 Department of Pharmacology and Molecular Sciences, Johns Hopkins University School of Medicine
5 McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine
6 Departments of Biomedical Engineering and Mental Health, Johns Hopkins University Schools of Medicine, Engineering, and Public Health
* These authors contributed equally to this work
+ Co-corresponding authors

The Genotype-Tissue Expression (GTEx) project established a data resource and tissue bank to study the relationship between genetic variation and gene expression in multiple human tissues, collecting whole-genome sequence and RNA-sequence from ~850 deceased adult donors. The Enhancing GTEx (eGTEx) project extends this resource by performing multiple molecular assays on tissue from GTEx donors [1].

As part of eGTEx, we have performed whole-genome bisulfite sequencing (WGBS) on 224 GTEx samples to measure DNA methylation and analyse its relationship with gene expression, genetic variation, and the other molecular marks assayed by eGTEx. Three-quarters of these samples come from 8 brain regions, reflecting the ambition of the eGTEx project to help contextualize results from GWAS of neurological and neuropsychiatric traits, and our group's interest in neuroepigenetics. Indeed, the eGTEx study design builds upon our recent work studying DNA methylation using WGBS from 72 human samples, covering 4 brain regions and 3 cell populations: nuclei positive for the neuronal marker NeuN (RBFOX3), negative for NeuN, and bulk tissue [2].

Here, we present results from [2] – including analyses of WGBS, ATAC-seq and RNA-seq – alongside the initial results for the 224 WGBS eGTEx samples. We identified tens-to-hundreds of megabases of differentially methylated regions (DMRs) and found that brain region-specific DMRs are almost only found in NeuN+ nuclei. These DMRs have characteristics of enhancers and are highly enriched for heritability of neuropsychiatric disease, such as schizophrenia, as estimated using genome-wide association study data.

Finally, we combined data from [2] with the eGTEx dataset to design capture probes (~46 Mb) for use with a bisulfite-sequencing assay targeting brain-specific DMRs, variably methylated regions (VMRs), and other brain-specific methylation loci. We are using this capture design to assay ~800 GTEx samples to explore how genetic variation influences DNA methylation in the adult human brain.

[1] https://doi.org/10.1038/ng.3969
[2] https://www.biorxiv.org/content/early/2017/03/24/120386

KEY WORDS
DNA methylation, neuroepigenetics, epigenetics, human, GTEx, eGTEx

2:30 PM - 2:50 PM

The Landscape of Circular RNA Expression In The Human Brain

Invited Abstract

Akira Gokoolparsadh
UNSW

BIOGRAPHY:

Akira Gokoolparsadh is a PhD student in Dr. Irina Voineagu's lab at UNSW working on the biogenesis and regulatory role of circular RNA in the human brain.

The Voineagu lab investigates the molecular genetic mechanisms underlying normal brain function and their perturbation in neurodevelopmental disorders including autism spectrum disorder.

ABSTRACT:

Akira Gokoolparsadh[1], Firoz Anwar[1], Irina Voineagu[1]

[1] School of Biotechnology and Biomolecular Sciences, University of New South Wales, Kensington, Sydney NSW 2052 Australia

Circular RNAs (circRNAs) are enriched in the mammalian brain and are upregulated in response to neuronal differentiation and depolarisation. These RNA molecules, formed by non-canonical back-splicing, have both regulatory and translational potential.

Despite accumulating evidence for a functional role of circRNAs in the brain, our current understanding of circRNA expression in the human brain is limited to a small number of samples.

Therefore, it remains unclear how circRNA expression varies across individuals, developmental stages and aging.Here, we generated the largest circRNA expression dataset available to date for the human brain, including nearly 200 human brain samples, from both healthy individuals and autism cases. We identify hundreds of novel circRNAs, characterise inter-individual variability of circRNA expression in the human brain, and identify brain region-specific circRNAs. We demonstrate that similarly to mRNAs, circRNA isoforms are not expressed stochastically, but rather as major isoforms, supporting their regulated expression in the human brain. We also find that circRNA expression is dynamic during brain development, decreasing with cellular maturation in brain organoids, but remains stable across the adult lifespan. These data provide a comprehensive catalogue of circRNAs and a deeper insight into their expression in the human brain.

KEY WORDS

Transcriptome, Non-coding RNAs, Circular RNAs, RNA-seq, Human Brain.

2:50 PM - 3:10 PM

Uncovering novel aspects of human neural crest biology using multiomic approaches

Invited Abstract

Dr Anushree Balachandran
University of Queensland

BIOGRAPHY:

Anushree has been working in the field of human stem cell biology for the last 7 years with a focus on disease modeling. She recently completed her doctoral studies at the Australian Institute of Bioengineering and Nanotechnology, The University of Queensland. The focus of her PhD project was to understand the molecular and functional profiles of human neural crest cells and their role in Down syndrome using genome editing, multiomic and bioinformatics approaches. The outcomes of her research include the identification of previously unexplored developmentally staggered populations of human neural crest cells, and their transcriptomes and proteomes, which reveal known and novel regulators of neural crest development as well as unique cell surface markers that enable their isolation. Her current interests lie in understanding early human developmental biology through the combination of pluripotent stem cell differentiation, genomics and bioinformatics.

ABSTRACT:

Anushree Balachandran[1], Dmitry Ovchinnikov[1], Keyur Dave[2], Jeffrey Gorman[2], Daniel Zalcenstein[3], Shian Su[3], Shalin Naik[3], Jarny Choi[4], Christine Wells[4], Samuel Zimmerman[5], Jessica Mar[1,5], Nick Glass[1] and Ernst Wolvetang[1]

[1] Australian Institute of Bioengineering and Nanotechnology, The University of Queensland, Australia
[2] QIMR Berghofer Medical Research Institute, Royal Brisbane and Women's Hospital, Australia
[3] Walter Eliza Hall Institute of Medical Research, Australia
[4] Centre for Stem Cell Systems, The University of Melbourne, Australia
[5] Systems and Computational Biology, Albert Einstein College of Medicine, New York, USA

Neural crest cells (NCCs) are a transient, multipotent population that arise at the neural plate border during embryonic development. They undergo epithelial to mesenchymal transition (EMT), delaminate from the dorsal neural tube and initiate the expression of SOX10 that establishes their identity as bona fide migratory NC.

Abnormalities in NC development lead to neurocristopathies such as Hirschsprung's disease, craniofacial and congenital heart defects. Infants born with Down syndrome (DS) have an increased risk of developing these, suggesting that trisomy 21 causes NC defects.

We developed a protocol to generate SOX10+ NCCs from human pluripotent stem cells. To obtain pure populations of SOX10+ NCCs, we employed CRISPR/Cas9 genome editing technology for site-specific insertion of a reporter cassette containing coding sequences for mMaple and Puromycin-N-acetyltransferase into the 3'UTR of the SOX10 locus. We show that reporter line derived SOX10:mMaple+ cells uniformly express SOX10 protein and are resistant to Puromycin, permitting their facile purification.

To gain insight into the molecular profiles of SOX10:mMaple positive and negative cells, we performed an in-depth multiomic characterisation study. Bulk RNA-seq revealed that SOX10+ cells are akin to cranial migratory NCCs, while SOX10- cells resemble specified premigratory NCCs. We further demonstrate that SOX10+ NCCs exhibit morphological and motility profiles consistent with an early migratory cranial NCC identity and are patternable with retinoic acid. Single cell RNA-seq of these NCCs revealed intra- and inter- population heterogeneity and potential causes thereof. Discovery proteomics further identified known and novel regulators of NC EMT, migration and morphology, and a cell surface marker specific to SOX10+ NCCs. This marker next permitted the isolation of pure SOX10+ cells from non-tagged wild type and DS-affected hPSCs, and the identification of proliferation defects of DS-NCCs. Collectively, our datasets contribute to a better understanding of NC developmental biology and exemplify a strategy for investigating human neurocristopathies.

KEY WORDS

Neural crest, Down syndrome, SOX10 reporter cell line, Multiomics,

3:10 PM - 3:30 PM

Big Data Technologies Enables Distant Relationship and Disease Variant Discovery in ALS.

Invited Abstract

Dr Natalie Twine
CSIRO

BIOGRAPHY:

Dr Natalie Twine is a postdoctoral fellow in the CSIRO transformational bioinformatics team and a visiting fellow at Macquarie University. Her research focuses on using genomic and transcriptomic data for translational and clinical impact. Dr Twine is passionate about using technology to improve health and understand complex disease. Dr Twine works collaboratively on a number of international and national projects, including the ALS Project MinE consortium and previously, as a visiting scientist at Southern Denmark University. Dr Twine has recently published in high impact journal, Neuron (Cell Press) and has 21 peer-reviewed publications with 944 citations and h-index of 12. She obtained her PhD in Bioinformatics from University of New South Wales and has previously worked at UNSW, Kings College London and University College London.

ABSTRACT:

Twine NA[1], Szul P[1], Williams KL[2], Dunne R[1], Blair IP[2], Bauer DC[1]

[1] CSIRO, Sydney, NSW, Australia
[2] Macquarie University, Sydney, NSW, Australia

Amyotrophic lateral sclerosis, (ALS) is a devastating neurodegenerative disorder, with no treatment, leading to death within 5 years. 10% of ALS cases are familial (FALS), while the remaining cases are sporadic (SALS). The genetic origins of ALS are poorly understood. FALS gene mutations are sometimes found in apparently SALS cases, indicating cryptic relatedness. Using large whole genome sequence (WGS) cohorts, we can identify unknown relationships, increasing the power to identify novel ALS genes. However, available relatedness tools are lacking in accuracy or speed. To illustrate the shortcomings of these tools we apply them to an ALS WGS cohort (n=838). The dataset contains known relationships (n=89). Using KING we were able to identify 97%, 84%, 82%, 59%, 40%, 24%, 13% and 9% of known 1st, 2nd, 3rd, 4th, 5th, 6th, 7th and 8th degree relatives respectively. While segment-based IBD tools (GERMLINE, ERSA) are more accurate at distant degrees, they are computationally slow hence not suitable for large WGS cohorts (Ramstetter, M D. et al.). To improve on speed and accuracy of existing tools, we are developing a relatedness tool as part of the VariantSpark framework (O'Brien, AR et. al.). We have simulated a 15-generation pedigree using WGS data with 831405 relationship pairs. Using ERSA with this simulated pedigree, we identified 100%, 85%, 84%, 67%, 54%, 41%, 34%, 24% and 21% of 1st, 2nd, 3rd, 4th, 5th, 6th, 7th, 8th and 9th degree respectively. We have already shown our VariantSpark relatedness tool to be 10x faster at processing the simulated pedigree than ERSA. We will also compare accuracy of ERSA with that of our relatedness tool. Furthermore, we will apply VariantSpark to allegedly unrelated SALS WGS samples (n=749) and report cryptic relatedness in this cohort. Finally, our tool will identify genomic segments shared IBD, thereby highlighting candidate ALS genes to be investigated.

KEY WORDS

Amyotrophic Lateral Sclerosis, Relatedness, Whole Genome Sequencing, Disease gene discovery, Big Data, Spark

SESSION 3: New Technologies Part 1

Chairs: Mark Van der Hoek & Vikki Marshall

4.30 PM - 5:00 PM

DNA Nanoballs and Single-Tube LFR: Affordable "Perfect" Genome Sequencing

Keynote Speaker

Dr. Rade Drmanac

Complete Genomics, Inc, a BGI/MGI company, California, US

BIOGRAPHY:

Dr Radoje (Rade) Drmanac, chief scientific officer and co-founder of Complete Genomics since 2006 and senior vice president of BGI since 2013, is a research scientist and inventor in the field of human genome sequencing including techniques such as DNA sequencing-by-hybridization (SBH), genomic micro- and nanoarrays, combinatorial probe ligation, and long fragment read (LFR) process for accurate whole genome sequencing and haplotyping from 10 human cells.

ABSTRACT:

Dr. Rade Drmanac

Complete Genomics, Inc, a BGI/MGI company, California, US

Precise personalized medicine depends on complete, accurate and phased WGS tests that are widely affordable. Our advanced massively parallel NGS platform based on the PCR-free error-free DNA nano-ball (DNB) nanoarrays (DNA spots <200nm), termed DNBseqTM, provides highly accurate pair-end 150-200 base WGS reads with extreme efficiency.

DNB nanoarrays are prepared by rolling circle replication in solution (Drmanac et al, Science, 2010) generating ~300 copies of the original DNA template without making copy of a copy.

DNB arrays are the most efficient patterned DNA arrays (<500nm pitch) enabling the lowest cost WGS. In addition, DNB arrays provide several fold higher template density per spot resulting in higher quality sequencing data than PCR-based cluster arrays that also suffer from amplification errors.

Recent improvements in making PCR-free WGS libraries and DNB arrays with ~500 base sequencing templates enable WGS with highest accuracy in detecting both SNPs and indels. These nanoarrays combined with novel fluidics and fast imaging cameras and scanning stages enable an order of magnitude more efficient generation of sequence data. We expect to be the first to achieve $100 genome.

Single-tube Long Fragment Read (LFR) technology that generates uniquely co-barcoded reads for each 30kb-300kb long genomic DNA fragment at the cost of regular libraries is another revolutionary genomic technology we recently developed (Nature 2012, bioRxiv 2018). It allows separate (phased) assembly of parental chromosome sequences.

Using up to 100 million barcodes from a pool of ~2 billion barcodes coupled to microbeads we uniquely barcode ~10 million long DNA fragments from 1ng of human DNA. Using co-barcoded reads, accurate WGS including detection and phasing of de novo mutations (~1 error per Gb, ~6 errors per genome) is achievable from ~10 cells (Genome Research 2015). In addition, single-tube LFR's uniquely co-barcoded reads obtained on DNBseqTM platform enable "perfect" (de novo assembled accurate and phased) personal WGS at affordable price.

5:00 PM - 5:20 PM

Reconstructing haplotyped methylomes with long-read sequencing for fine dissection of imprinting in mice

Invited Abstract

Dr Quentin Gouil
Walter And Eliza Hall Institute

BIOGRAPHY:

I am interested in the epigenetic mechanisms of gene regulation in plants and animals.

ABSTRACT:

Scott Gigante[1,6*†], **Quentin Gouil[2,3†]**, Alexis Lucattini[4†], Andrew Keniry[2,3], Tamara Beck[2], Matthew Tinning[4], Lavinia Gordon[4], Chris Woodruff[1], Terence P. Speed[1,3^], Marnie E. Blewitt[2,3^] and Matthew E. Ritchie[2,3,5^]

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† Equal contribution
^ Equal contribution

Asymmetric expression patterns between the two parental alleles are critical for development of the mammalian embryo. This process known as imprinting involves differential DNA methylation of the parental genomes. We sequence mouse embryonic placental tissue from reciprocal crosses on the Oxford Nanopore MinION and PromethION platforms, and exploit the long reads to determine both haplotype and CpG methylation levels. Comparison with matched Reduced-Representation Bisulfite Sequencing data confirms the accuracy of the methylation calls, and highlights the improvement in haplotyping conferred by the longer reads.

We successfully identify known imprinting control regions and refine their genomic coordinates. Combining de novo differential methylation calling with allele-specific RNA-seq also identifies new correlations between monoallelically expressed genes and allele-specific methylation. The approach we developed to reconstruct haploytped methylome is simpler, cheaper and more powerful than short-read techniques, allowing a greater resolution in the investigation of epigenetic regulation.

KEYWORDS

nanopore sequencing; DNA methylation; imprinting; haplotyping

5:20 PM - 5:40 PM

Reconstructing haplotyped methylomes with long-read sequencing for fine dissection of imprinting in mice

Invited Abstract

Dr Martin Smith
Garvan Institute of Medical Research

BIOGRAPHY:

Dr Smith majored in microbiology and immunology before obtaining a Master's degree in Bioinformatics at the University of Montreal. He then moved to Brisbane to pursue a PhD in Genomic and Computational Biology with Prof John Mattick, aiming to find new functional regions in the non-coding genome. He joined the Garvan Institute as a research officer in 2012 and now leads the Genomic Technologies program at the Kinghorn Centre for Clinical Genomics, where his team develops new genomic applications for translational research and new clinical tests.

ABSTRACT:

Ghamdan Al-Eryani[1,2], Mandeep Singh[1,2], Shaun Carswell[1,2], Katherine Jackson[1,2], James M. Ferguson[1,2], Kirston Barton[1,2], Daniel Roden[1,2], Sunny Z. Wu[1,2], Chia-Ling Chan[1,2], David Koppstein[2,3], Fabio Luciani[2,3], Tri Phan[1,2], Joseph Powell[1,2], Simon Junankar[1,2], Chris C. Goodnow[1,2], ***Martin A. Smith[1,2]**, *Alex Swarbrick[1,2]

[1] Garvan Institute of Medical Research, Darlinghurst, NSW, Australia
[2] St Vincent's Clinical School, Faculty of Medicine, UNSW Sydney, Randwick, NSW, Australia
[3] Kirby Institute for Infection and Immunity, UNSW Sydney, Randwick, NSW, Australia

High throughput single cell RNA sequencing (scRNAseq) is providing revolutionary insights into biological systems and disease. Most scRNAseq protocols are designed for short read sequencing and, thus, only produce information from one end of cDNA molecules. This is often sufficient for transcriptome profiling and 3' or 5' end annotation, but restricts other research applications including splicing, lymphocyte receptor repertoire, or somatic variation studies. Long-read sequencing can overcome these limitations, but the higher per-base error rate impedes detection of cell barcode and unique molecular indices (UMIs), which are essential to deconvolute scRNAseq data.

We have developed RAGEseq (Receptor And Gene Expression sequencing) to overcome these challenges. After partitioning thousands of single cells with the 10X Chromium platform, cDNA libraries are split and undergo targeted hybridization capture followed by Oxford Nanopore sequencing. Libraries are analyzed in parallel with Illumina sequencing to perform transcriptome profiling and accurately resolve cell barcodes and UMIs. The short read data is then used to demultiplex the long read data, which are in turn used to resolve the sequence of full-length transcripts via de novo assembly, error correction, and polishing.

After benchmarking on cell lines and peripheral blood monocytes, we used RAGEseq to profile the transcriptomes of cells sampled from the primary tumour and draining lymph node of a breast cancer patient, including full-length T-cell (TCR) and B-cell (BCR) receptor sequences. We use RAGEseq to phenotype and track clonally related lymphocytes between tissues,

identify BCR transcripts encoding antibodies destined for secretion versus membrane localization, observe the evolution of somatic hypermutation in B-cells, and reveal the expression of novel TCR transcripts.

KEY WORDS

Single cell sequencing, RNAseq, Oxford Nanopore, 10X Chromium, Targeted sequencing, Cancer, Immunology

5:40 PM - 6:00 PM

Cool things that can be done with Optical Mapping

Invited Abstract

Dr Eva KF Chan
Garvan Institute of Medical Research

BIOGRAPHY:

Dr Eva Chan completed her PhD in Bioinformatics at the University of New South Wales, where she studied the Genetic Influence of Gene Expression. Following two Bioinformatics postdoctoral positions, at CSIRO (QLD, Australia) and University of California, Davis (CA, USA), Dr Chan took up a position as the Statistical Genetics Lead at the Vegetable Seeds Division of Monsanto Company, in California, in 2009.

Returning to Sydney in 2013, Dr Chan joined the Garvan Institute of Medical Research, undertaking research in human diversity with particular interest in large-scale structural variations.

ABSTRACT:

Eva K F Chan[1,2], Ruth J Lyons[1], Vanessa M Hayes[1,2,3,4]

[1] Genomics and Epigenetics Division, Garvan Institute of Medical Research, NSW 2010, Australia
[2] St Vincent's Clinical School, University of New South Wales, NSW 2010, Australia
[3] Central Clinical School, University of Sydney, NSW 2006, Australia
[4] School of Health Systems and Public Health, University of Pretoria, Pretoria 0028, South Africa

Optical mapping is a 20-year-old molecular approach for constructing genome maps. Next-generation mapping is a modern advancement to this technique, with improvements including: (1) the obviated need for restriction enzymes, (2) ability to assay long DNA molecules (20 kb to megabase length), (3) ability to flow native double-stranded DNA through nano-channels rather than immobilizing them on slides, and (4) improvements in automation allowing high-throughput capture of millions of single molecules per experiment.

Recent years have seen successful application of this technology for scaffolding large genome assemblies and detection of both inherited (germline) and acquired (somatic) complex genomic rearrangements. We present examples of our utilization, from generating the first dingo reference genome, to the detection of multiple genomic rearrangements in matched-normal cancer samples, to identifying structural variants in a familial disease.

Novel ideas for unique applications of optical mapping are continually being developed to address biological questions in new ways, such as (1) optical methylation mapping to provide simultaneous information on structural variation and methylation status, (2) optical replication mapping to identify origins of replication in complex genomes such as human, and (3) enriched targeted optical mapping (without amplification) through the use of Cas9 excision.

How can optical mapping empower your research?

KEY WORDS

Optical Mapping, Genomics, Genome Assembly, Structural Variants

SESSION 4:
New Technologies Part 2

Chairs: Mark Van der Hoek & Vikki Marshall

9:00 AM - 9:45 AM

CITE-seq: Highly multiplexed simultaneous measurement of epitopes and transcriptomes in single cells

Keynote Speaker

Marlon Stoeckius,
New York Genome Center

BIOGRAPHY:

Dr. Marlon Stoeckius is a senior research scientist in the NYGC's technology innovation lab, a multidisciplinary research team, comprised of molecular biologists, engineers and chemists, seeking to advance a variety of next-generation sequencing techniques. His major focus is the development of experimental methods to obtain multimodal information from single cells. Prior to joining NYGC, he worked as postdoctoral researcher at Yale University studying gene expression regulation in early embryogenesis. He performed his graduate research in the lab of Nikolaus Rajewsky at the Max-Delbrueck-Center for Molecular Medicine Berlin, characterizing the oocyte-to-embryo transition in C. elegans. Being part of an international MDC-NYU PhD exchange program, he did stints of his PhD research in the lab of Fabio Piano at New York University. He received his PhD in molecular biology from the Humboldt University Berlin, and his MA in biomedical sciences from the University of Applied Sciences Bonn, Germany.

ABSTRACT:

Marlon Stoeckius[1], Shiwei Zheng[2], Christoph Hafemeister[2], William Stephenson[1], Brian Houck-Loomis[1], Pratip K. Chattopadhyay[3], Bertrand Yeung[4], Harold Swerdlow[1], Rahul Satija[2], Peter Smibert[1]

[1] Technology Innovation Lab, New York Genome Center, New York, NY
[2] New York Genome Center, New York, NY; New York University Center for Genomics and Systems Biology, New York, NY
[3] New York University Medical Center, New York, NY
[4] BioLegend Inc., San Diego, CA

The last few years have seen the scale of single cell RNA-seq experiments increase exponentially greatly enhancing our understanding of cell biology in development and disease. It is now feasible for researchers to characterize thousands of single cells in one experiment. However, important hallmarks of immune cell states are often not detected in scRNA-seq experiments. While lower throughput methods previously allowed researchers to link phenotypes or protein expression to transcriptomic profiles, the increase in scale of modern droplet-based methods resulted in a loss of such addressability.
Here we describe two recently developed applications that utilize antibody conjugated oligos to enhance existing scRNA-seq platforms. 1. CITE-seq, which allows measurement of a potentially unlimited number of protein markers in parallel to transcriptomes. We applied CITE-seq to deeply profile the human immune system. We present an in-depth characterization of T cell populations in the human immune system using a panel of ~80 antibodies revealing complexity not previously observed by scRNA-seq. 2. Cell Hashing, which enables sample multiplexing, robust multiplet detection and super-loading of scRNA-seq platforms, allowing confident recovery of 4 times as many single cells per experiment.

9:45 AM - 10:10 AM

Dissecting plant development at single cell resolution

Invited Speaker
Professor Ryan Lister
University of Western Australia

BIOGRAPHY:

Ryan Lister leads a research group investigating the epigenome and cell identity, at the University of Western Australia and the Harry Perkins Institute of Medical Research. After receiving his PhD from UWA in 2005, Ryan undertook postdoctoral studies at The Salk Institute for Biological Studies, where he developed and applied new techniques to map the epigenome and transcriptome. Having returned to UWA in 2012, Ryan's laboratory is focused upon understanding how the epigenome patterns are established and changed, how they affect the readout of underlying genetic information, their involvement in development and disease, and developing molecular tools to precisely edit the epigenome.

ABSTRACT:

Marina Olivia, Dave Tang, Tim Stuart, Jahnvi Pflueger, **Ryan Lister**

University of Western Australia, Perth WA, Australia

Challenges in the isolation of specific plant cell types and the inability to effectively culture plant cells of distinct defined identities have impeded the deep molecular characterization of the diversity of plant cell identities and developmental transitions. Moreover, the rigid cell wall that surrounds plant cells forms a challenging barrier to single cell dissociation and analysis. Consequently, little is known about the transcriptional state of very rare or transient cell populations. We have undertaken initial efforts at characterization of plant transcriptomes at single cell resolution through a combination of genomic and microscopy techniques. Improved experimental and computational methodologies for improved cell discrimination has enabled sensitive detection and mapping of transcriptional states of complex tissues, including rare, transient, and previously uncharacterized cell states. Reconstruction of developmental trajectories for distinct cell lineages revealed more complex topologies than expected. Finally, through analysis of the dynamics of gene expression we have gained new insights into the different gene regulatory mechanisms at play during plant cell differentiation.

SESSION 5:
Cancer

Chairs: Deb White & Vanessa Hayes

10:40 AM - 11:25 AM

From Petabytes to Molecular Insights: Building a System for Precision Medicine

Keynote Speaker
Dr Kevin White
University of Chicago

BIOGRAPHY:

Dr. Kevin White serves as President at Tempus, where he oversees the scientific operation. Dr. White was the founding Director of the Institute for Genomics and Systems Biology at The University of Chicago, where he retains an appointment as the James and Karen Frank Family Professor of Human Genetics and Medicine. Dr. White is a pioneer in combining experimental and computational techniques to understand the networks of factors that control biological systems during development, disease, and evolution. His team has discovered novel cancer genes in a wide range of tumor types including breast cancer, kidney cancer and myeloid leukemias. From 2006 through 2016, while he was the Founding Director of the Institute for Genomics & Systems Biology, Dr. White built a team of

10 faculty and over 100 staff that has won approximately $150M in research funding contracts and awards. His academic awards for innovation include the Keck Biomedical Investigator and Beckman Young Investigator awards, and he has also been recognized for his entrepreneurship on the '40 under 40' list by Chicago Crain's Business. White has served on the scientific boards of a wide range of start-up biotech companies and foundations. He also has previously served as acting CEO and co-director of HealthSeq Pvt. Ltd., providing hospitals with protected data clouds for analysis of genome and clinical records data. In the nonprofit sector he currently holds scientific advisory board positions at institutions that include Harvard University, Yale University, Northwestern University and the National Cancer Institute. Dr. White has a B.S. and M.S. from Yale and a Ph.D. from Stanford University.

ABSTRACT:

Kevin White[1] [2]

[1] Tempus Labs, Chicago, Illinois USA
[2] University of Chicago, Chicago, Illinois USA

Over the last decade there has been a confluence of technological advances in the computational and biological sciences that set the stage for large-scale improvements in how cancers are diagnosed and treated, leading to a new vision for advancing precision oncology. As part of this vision we have taken steps to develop technological solutions and novel approaches to the challenge of integrating massive datasets and validating discoveries. In this context I will describe a series of proof-of-principle projects that our team has made to highlight basic discoveries and applications in cancer diagnostics and therapeutics. With these and many other efforts as a backdrop for what can be done, I will also describe our recent efforts at Tempus Labs to engineer and industrialize a systematic solution that is allowing physicians to treat patients enabled by a real-time learning system that condenses petabytes of data into actionable information matched uniquely to each patient.

11:25 AM - 11:45 AM

The super enhancer-driven long noncoding RNA lncNB promotes neuroblastoma tumorigenesis

Invited Abstract

Miss Sujanna Mondal
Childrens Cancer Institute Australia, Randwick, Australia

BIOGRAPHY:

Sujanna is a second year PhD student at the Children's Cancer Institute. She completed her Honours in neuroblastoma research under the supervision of A/Prof Tao Liu in the Histone Modification Group. Being highly passionate about childhood cancer she has continued her PhD looking into alternate therapeutic targets for this extremely prevalent disease.

ABSTRACT:

S.Mondal, M.Wong, A.Tee, P.Liu, and T.Liu

Children's Cancer Institute Australia

MYCN oncogene amplification in tumour tissues is a well-documented prognostic marker in high risk neuroblastoma patients, however 70% of high risk neuroblastomas do not exhibit this amplification. The expression of oncogenes associated with transcriptional super-enhancers are regulated by the BET bromodomain protein BRD4 and the transcription factor TFIIH-associated kinase CDK7. Altered expression of long noncoding RNAs (lncRNAs) in several cancers including neuroblastomas have been shown to function as diagnostic markers.

Here we performed RNA sequencing analysis of human neuroblastoma cell lines and tumour tissues. The expression of the novel lncRNA lncNB was found to be positively associated with c-Myc expression in MYCN non-amplified neuroblastoma cell lines and patient tumour tissues. High expression of lncNB in tumour tissues was also associated with poor patient prognosis. Bioinformatics analysis of published histone H3K27 acetylation chromatin immunoprecipitation sequencing data

revealed super-enhancers at the lncNB gene locus only in MYCN non-amplified cells.

Next, we examined the role of lncNB in neuroblastoma. Transfection of MYCN non-amplified neuroblastoma cell lines with siRNAs targeting the super-enhancer regulator BRD4 over 48 hours resulted in decreased lncNB expression. Treatment with the small molecular BRD4 inhibitor AZD5153 or the CDK7 inhibitor THZ1 reduced cell proliferation and lncNB expression in MYCN non-amplified neuroblastoma cells. Knockdown of lncNB with siRNAs or doxycycline-inducible shRNAs in MYCN non-amplified cell lines led to a decrease in cell viability over 96 hours as well as downregulation of c-Myc protein expression within 48 hours. Furthermore, doxycycline-induced knockdown of lncNB in MYCN non-amplified shRNA cell lines led to a significant decrease in colony formation over 14 days.

This data suggests that the lncRNA lncNB is upregulated by super-enhancer activity in MYCN non-amplified neuroblastoma cell lines, and that up-regulation of lncNB induces neuroblastoma cell proliferation, survival and clonogenic capacity. Targeting lncNB expression can be an effective therapeutic strategy.

KEY WORDS

Neuroblastoma, super-enhancer, lncNB, MYCN non-amplified

11:45 AM - 12:05 PM

MicroRNAs regulate large-scale alternative splicing in cancer by controlling the RNA-binding protein Quaking.

Invited Abstract

Dr Katherine Pillman
University of South Australia

BIOGRAPHY:

Dr Pillman is a bioinformatician with a broad interest in many areas of gene regulation. Originally a wet-bench biologist, she began by exploring regulation of gene networks in barley (Australia) and potato plants using RNA-seq (Oregon State University), which fuelled her interest in bioinformatics. In 2012, she took up the role of lead bioinformatician with Prof Greg Goodall, working on gene regulatory mechanisms and networks in cancer with a focus on epithelial-mesenchymal transition (EMT). Highlights include identifying Quaking as the first known protein to control the formation of circular RNAs and characterising circular RNAs during EMT (Cell 2015) and characterising the role of Quaking in mRNA alternative splicing in cancer (EMBO J, 2018). Her current work involves using a range of genomics next-generation sequencing data types to dissect gene regulation, including analysis of alternative splicing, microRNA biology and targeting, circular RNAs, epigenetic modifications, gene regulatory networks and expression.

ABSTRACT:

Pillman, K.A.*[1,2],, Phillips, C.A.*[1], Toubia, J.*[1,2], Roslan, S.*[1], Dredge K.[1], Bert A.G.[1], Khew-Goodall Y.[1,2], Selth L.A.6, Goodall, G.J.[1,2] and Gregory, P.A.[1,2]

[1] Centre for Cancer Biology, University of South Australia and SA Pathology, Australia
[2] University of Adelaide, Australia
[3] Dame Roma Mitchell Cancer Research Laboratories, University of Adelaide, Australia

MicroRNAs are critical gatekeepers of the epithelial cell state, restraining expression of pro-mesenchymal genes that drive epithelial-mesenchymal transition (EMT) and contribute to metastatic cancer progression. We have discovered that through strong suppression of the RNA-binding protein Quaking, miR-200c and miR-375 also exert widespread control of alternative splicing patterns in cancer cells.

Deep RNA sequencing and HITS-CLIP revealed that Quaking binds to and directly regulates hundreds of EMT alternative splicing events without appreciably affecting gene expression levels. Quaking is both necessary and sufficient to drive splicing changes and controls key cancer-related cell properties such as migration and invasion. Analysis of The Cancer Genome Atlas data shows that the QKI-driven splicing signature is broadly conserved across many cancer types, further indicating its functional importance. Quaking-driven splicing targets are enriched within the actin cytoskeleton regulatory network, where the functions of Quaking and miR-200 converge and several genes are directly targeted by both Quaking and miR-200c, revealing coordinated control of alternative splicing and mRNA abundance during EMT.

These findings demonstrate that a miR-200/Quaking axis controls alternative splicing and has a critical impact on cancer-associated epithelial cell plasticity.

KEY WORDS

bioinformatics, alternative splicing, gene expression, microRNA, RNA-seq, HITS-CLIP, cancer.

KEY WORDS →

Cancer genomics, tumour development, mutation signatures, cancer treatment

1:15 PM - 1:40 PM

Whole Genome Sequencing of Tumour and Normal Samples to Identify Cancer Aetiology and Therapeutic Opportunity

Invited Speaker

Dr Nic Waddell

QIMR Berghofer Medical Research Institute

BIOGRAPHY:

Nic WADDELL is head of the Medical Genomics group at QIMR Berghofer Medical Research Institute, Australia. She is an NHMRC senior research fellow and a cancer researcher who uses bioinformatics to interpret next generation sequence data. She is involved in several genomics projects into a variety of cancer types including familial breast cancer, melanoma and mesothelioma. Her research focuses on the identification of mutational processes and therapeutic opportunity in cancer, and she has a passion for seeing genomics implemented into the clinic.

ABSTRACT:

Nic Waddell[1]

[1] QIMR Berghofer Medical Research Institute, Brisbane

Genome sequencing has made great advances in our understanding of tumour development and progression. Cancer genome data is being used to classify tumours into significant subtypes, discover driver mutations, identify the mutation processes that underlie tumour development and find alternative therapeutic targets. These are important steps towards 'personalised medicine' where the diagnosis, management and treatment of patients are based on an individual's clinical and genomic data. We have been working on a variety of cancer types including familial breast cancer, melanoma and mesothelioma. This talk will describe some of our recent findings. There will be a particular emphasis on the potential clinical utility of cancer sequencing, including i) the identification of germline predisposition variants, and ii) the identification of candidate markers of treatment including immunotherapy.

1:40 PM - 2:00 PM

Landscape of the breast cancer tumour microenvironment using single-cell RNA sequencing

Invited Abstract

Sunny Z. Wu
Garvan Institute of Medical Research

BIOGRAPHY:

Sunny is a PhD Student in A/Prof. Alex Swarbrick's group at the Garvan Institute of Medical Research who is exploring the tumour microenvironment in breast cancer using emerging technologies in single-cell genomics.

ABSTRACT:

Sunny Z. Wu[1,2], Daniel Roden[1,2], Ghamdan Al-Eryani[1,2], Chia-Ling Chan[1], Kate Harvey[1], Holly Holliday[1,2], Rui Hou[5], Mun Hui [1,2,3], Davendra Segara[4], Andrew Parker[4], Sanjay Warrier[4], Cindy Mak[3,6], Alistair Forrest[5], Nenad Bartonicek[1,2], Elgene Lim[1,2,4], Sandra O'Toole[1,2,7], Simon Junankar[1,2], Aurélie Cazet[1,2], and Alexander Swarbrick[1,2]

1 The Kinghorn Cancer Centre and Cancer Research Division, Garvan Institute of Medical Research
2 St Vincent's Clinical School, Faculty of Medicine, University of New South Wales
3 The Chris O' Brien Lifehouse
4 St Vincent's Hospital
5 Harry Perkins Institute of Medical Research, University of Western Australia
6 Royal Prince Alfred Hospital
7 Australian Clinical Labs

Breast cancers are a complex 'ecosystem' of diverse cell types, whose heterotypic interactions between malignant, stromal and immune populations are central in defining the aetiology of the disease and response to therapy. Despite advances in other carcinomas, including immune-checkpoint blockade in melanoma, combinational therapies that target the supporting microenvironment have made little clinical progress in breast cancer treatment. The development and implementation of such therapies has been largely impeded by a poor understanding of the cellular heterogeneity within breast cancers, which is masked using conventional bulk sequencing approaches.

Single-cell RNA sequencing (scRNA-Seq) technologies and computational methods have emerged as remarkable tools for studying the diverse cellular populations within the tumour microenvironment. Using this approach, we comprehensively profiled more than 120,000 neoplastic, immune and parenchymal cells sampled from 22 primary and metastatic breast cancers collected at surgery and biopsies. At single-cell resolution, we describe novel immune and stromal subsets and infer their intra-tumour interactions, leading to important mechanistic insights with therapeutic implications. Using the CITE-SEQ method, we provide simultaneous cell-surface protein levels with single cell transcriptomics to comprehensively phenotype novel immune subsets and redefine their transcriptional signatures, guiding new strategies for immunotherapy in breast cancer. In addition, we profiled the heterogeneity of cancer-associated fibroblasts (CAFs) across primary and metastatic sites and propose multi-faceted roles in regulating malignancy and tumour immunology. Using co-expression and motif enrichment, we predict distinct transcription factor networks regulate these polarised states. Novel cell surface markers identified using scRNA-Seq allow us to prospectively isolate CAF subsets for the validation of targetable gene expression features.

This is by far the largest and most comprehensive single cell genomic study in any cancer to date. Our approach highlights the power of single cell technologies to unravel the complexities of the tumour microenvironment and identify novel mechanisms underlying carcinogenesis. Such insights will guide the next-generation of therapies, which will likely be based upon an integrated understanding of the neoplastic, stromal and immune states that define a tumour and inform treatment response in breast cancer.

KEY WORDS

Single Cell RNA Sequencing; Breast Cancer; Tumour Microenvironment; CITE-SEQ

2:00 PM - 2:20 PM

Going nuclear: Using single-nuclei RNA-sequencing for analysis of rare tumours

Invited Abstract

Dr Richard Tothill
Peter MacCallum Cancer Centre

BIOGRAPHY:

I completed my PhD through the Department of Pathology at University of Melbourne and at the Peter MacCallum Cancer Centre. I have 20 years of experience in both academic research and the pharmaceutical industry and currently lead the Rare Disease Oncogenomics Laboratory at the University of Melbourne Centre for Cancer Research (UMCCR). My current and past research encompasses genomic landscape analyses of solid adult cancers incorporating the methods of DNA sequencing and gene-expression profiling with an emphasis on clinical translation and development of diagnostic methods. My past highly cited work has involved molecular classification of solid tumours such as ovarian cancer and cancers of unknown primary. More recently I have focused on understanding rare and cancers including neuroendocrine tumours (NETS).

ABSTRACT:

Luciano Martelloto[1], Magnus Zethoven[2], **Richard Tothill[1,2]**

[1] Centre for Cancer Research and Department of Clinical Pathology, University of Melbourne
[2] Peter MacCallum Cancer Centre

Single cell RNA-sequencing is a transformative new technology for understanding cancer biology. Tissue dissociation and then transcriptional profiling of single cells can uncover the complex cellular makeup of individual tumours that can often be lost by bulk gene-expression analysis. However, the logistics of processing and then preserving human tissue samples that are then compatible with single cell methods is not trivial. In rare cancers these issues are further compounded by the infrequent opportunities to bank such tumours. Single nuclei RNA-seq (sn-RNA-seq) is an alternative approach that enables the analysis of snap frozen tissue samples, allowing the analysis of large collections of frozen samples that have been banked over many years. We have optimised sn-RNA-seq on the 10x chromium platform and applied this to the single cell analysis of rare endocrine tumours. Phaeochromocytomas and paragangliomas (collectively PCPG) are remarkable for their hormone-producing functionality, high heritability and genetic diversity. By bulk gene-expression profiling these tumours can be clustered to four to six distinct gene-expression subtypes. Gene expression signatures associated with some subtypes indicate an abundance of stromal and immune cells. For example, pseudohypoxic tumours, associated with mutations in the tumour suppressor gene VHL, are highly vascularized and contain a large number of endothelial cells. Some PPGL tumours also exhibit high expression of immune cell markers indicating an abundance of lymphocytes or macrophages. We used sn-RNA-seq of snap frozen PCPG tumours previously characterised using bulk RNA-seq. We identified a diversity of cell types at unprecedented resolution in these tumours that has allowed us to better understand the cellular composition or PCPG gene-expression subtypes and tumour cell specific profiles. We have furthermore shown that sn-RNA-seq can be used to identify copy-number variation and clonal heterogeneity between tumour cell populations. Single-nuclei RNA-seq is therefore a breakthrough method for the analysis of rare cancers.

KEY WORDS

Single cell, single nuclei, transcription, rare cancer

SESSION 6: Bioinformatics

Chairs: Andreas Schreiber & Kat Pillman

3:20 PM - 4:05 PM

Genomic approaches to decipher the extent and impacts of X chromosome inactivation

Keynote Speaker

Dr Taru Tukiainen

Institute for Molecular Medicine Finland

BIOGRAPHY:

Dr Tukiainen is a newly appointed group leader at the Institute for Molecular Medicine Finland, FIMM, at the University of Helsinki, funded by the Academy of Finland Research Fellowship. Her work focuses on understanding the genomic underpinnings of sex differences in health and disease. Taru received a D.Sc. (Tech.) degree in Computational Systems Biology in 2012 from Aalto University, Finland, after which she conducted a postdoc in Daniel MacArthur's research team at the Massachusetts General Hospital and the Broad Institute prior returning to Finland and FIMM in 2015.

ABSTRACT:

Taru Tukiainen[1]

[1] Institute for Molecular Medicine Finland, Helsinki, Finland

X chromosome inactivation (XCI) serves to balance the X-chromosomal dosage between XX females and XY males by silencing the transcription from one of the two X chromosomes in female cells. The silencing by XCI is, however, largely incomplete, as first established more than a decade ago: up to one third of X chromosome genes escape from XCI, i.e., remain expressed from both X chromosomes in females to some degree.

This incompleteness in XCI may be a contributor in male-female differences, yet given technological limitations and lack of suitable data sets, few comprehensive studies on XCI and its downstream effects in human samples have been conducted. To systematically survey the landscape of XCI across human tissues, we have analyzed over 5,500 transcriptomes from 449 individuals spanning 29 tissues from the GTEx data set together with ~1000 single-cell transcriptomes combined with genomic sequence data. In line with previous work, we find that incomplete XCI affects at least 23% of X-chromosomal genes. Given the unprecedented resolution and breadth of our data, we further demonstrate that XCI is generally uniform across human tissues, but with notable examples of heterogeneity between tissues, individuals and cells, and provide insights into the sources of variability in XCI. Our data also shows that a large majority of escape genes show male-female expression differences detectable at population-level, thus highlighting the potential between-sex diversity introduced by incomplete XCI. In our current work we are extending these findings to human complex traits and I will show examples of how incomplete XCI can manifest as sex differences in phenotypic traits. Overall, this work on human XCI, facilitated by the deployment of several complementary genomic approaches, helps to increase our understanding of the extent and impacts of the incompleteness of XCI.

KEY WORDS

X chromosome inactivation, XCI, sex differences, transcriptomics, single cell sequencing, RNA sequencing, complex traits, GTEx, epigenetics

4:05 PM – 4:30 PM

Form versus Function: Understanding the beauty in genomic noise

Invited Speaker
Nicole Cloonan
The University of Auckland

BIOGRAPHY:

Nicole Cloonan is an Associate Professor in Bioinformatics at The University of Auckland. She was previously an ARC Future Fellow at the QIMR Berghofer Medical Research Institute, and an ARC Postdoctoral Fellow at The University of Queensland. Her work is multi-disciplinary in nature, involving computational biology and bioinformatics, biochemistry, cell biology, and molecular biology – all of which she uses to understand the complexity of RNA systems. She's pretty awesome, you should come to New Zealand and do a PhD with her.

ABSTRACT:

Nicole Cloonan[1]

[1] The University of Auckland

Genome biology commands tremendous public, commercial, and political interest, as it holds great promise for solving many of humanities biggest challenges. However, our understanding of genomes has not kept pace with our ability to measure them, and in the rush to discover and classify and catalogue and publish in a hyper-competitive environment we are often guilty of failing to fully test our assumptions or clarify our definitions. Determining what regions of our genome are functional is a core goal of most genomic researchers, but separating signal from noise can be problematic, and has led to dogmatic definitions that often repeated but rarely justified. This talk proposes that in order to understand function, you have to also understand form, or to put it another way, in order to understand signal, you have to also understand noise.

4:30 PM - 4:50 PM

STRetch: detecting and discovering pathogenic short tandem repeat expansions

Invited Abstract
Mrs Harriet Dashnow
Murdoch Children's Research Institute

BIOGRAPHY:

Harriet Dashnow is a bioinformatician and PhD candidate at the University of Melbourne and the Murdoch Children's Research Institute (MCRI). She has previously worked as a bioinformatician in the Department of Biochemistry at the University of Melbourne and the Victorian Life Sciences Computation Initiative (VLSCI), where she worked on the Melbourne Genomics Health Alliance project.

Harriet obtained a Bachelor of Arts (Psychology), a Bachelor of Science (Genetics and Biochemistry) and a Master of Science (Bioinformatics) with Dean's Honours from the University of Melbourne. She was awarded an Australian Postgraduate Award (APA), MCRI Top Up scholarship and the Australian Genomics Health Alliance Top Up scholarship for her PhD studies.

ABSTRACT:

Harriet Dashnow[1,2], Monkol Lek[3,4], Belinda Phipson[1], Andreas Halman[1,5], Simon Sadedin[1], Andrew Lonsdale[1], Mark Davis[6], Phillipa Lamont[7], Joshua S. Clayton[8], Nigel G. Laing[8], Daniel G. MacArthur[3,4] and Alicia Oshlack[1,2]

[1] Murdoch Childrens Research Institute, Royal Children's Hospital, Parkville, VIC, Australia
[2] School of Biosciences, The University of Melbourne, Parkville, VIC, Australia
[3] Analytic and Translational Genetics Unit, Massachusetts General Hospital, Boston, Massachusetts, USA
[4] Program in Medical and Population Genetics, Broad Institute of MIT and Harvard, Cambridge, Massachusetts, USA
[5] Florey Institute of Neuroscience and Mental Health, University of Melbourne, Parkville, VIC, Australia
[6] Department of Diagnostic Genomics, PathWest Laboratory Medicine, QEII Medical Centre, Nedlands, WA, Australia
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[8] Harry Perkins Institute of Medical Research, Centre for Medical Research, University of Western Australia, Nedlands, WA, Australia.

Short tandem repeat (STR) expansions have been identified as the causal DNA mutation in dozens of Mendelian diseases. Historically, pathogenic STR expansions could only be detected by single locus techniques, such as PCR and electrophoresis. The ability to use short read sequencing data to screen for STR expansions has the potential to reduce both the time and cost to reaching diagnosis and enable the discovery of new causal STR loci. Most existing tools detect STR variation within the read length, and so are unable to detect the majority of pathogenic expansions. Those tools that can detect large expansions are limited to a set of known disease loci and as yet no new disease causing STR expansions have been identified with high-throughput sequencing technologies.

Here we address this by presenting STRetch, a new genome-wide method to detect STR expansions at all loci across the human genome. We demonstrate the use of STRetch for detecting pathogenic STR expansions in short-read whole genome sequencing data with a very low false discovery rate. We further demonstrate the application of STRetch to solve cases of patients with undiagnosed disease and apply STRetch to the analysis of 97 whole genomes to reveal variation at STR loci. STRetch assesses expansions at all STR loci in the genome and allows screening for novel disease-causing STRs.

STRetch is open source software, available from github.com/Oshlack/STRetch.

KEY WORDS

Short tandem repeats (STRs), bioinformatics, human disease genomics

4:50 PM - 5:10 PM

Integrated pathogen load and dual transcriptome analysis of systemic host-pathogen interactions in severe malaria

Invited Abstract

Dr Hyun Jae Lee
QIMR Berghofer Medical Research Institute

BIOGRAPHY:

Hyun Jae Lee obtained his PhD from the Institute for Molecular Bioscience, University of Queensland, in 2017, under the supervision of Associate Professor Lachlan Coin, for research on understanding disease pathogenesis through RNA sequencing. Throughout his Ph.D., he worked on various aspects of transcriptomics, including noncoding RNAs, which involved analysis of big data and implementation of bioinformatics tools. He is currently working as a research officer at QIMR Berghofer Medical Research Institute working between two groups, focusing on using the single-cell data to study cancer and malaria.

ABSTRACT:

Hyun Jae Lee[1], Athina Georgiadou[2], Michael Walther[3], Davis Nwakanma[3], Lindsay B Stewart[4], Michael Levin[2], Thomas Otto[5], David J Conway[4], Lachlan J Coin[6], Aubrey J Cunnington[2]

[1] QIMR Berghofer Medical Research Institute, Brisbane, QLD, Australia
[2] Section of Paediatrics, Imperial College, London, UK
[3] Medical Research Council Gambia Unit, Fajara, The Gambia
[4] Department of Pathogen Molecular Biology, London School of Hygiene and Tropical Medicine, London, UK
[5] Wellcome Trust Sanger Centre, Hinxton, UK
[6] Institute for Molecular Bioscience, The University of Queensland, Brisbane, Australia

The pathogenesis of severe Plasmodium falciparum malaria is incompletely understood. Since the pathogenic stage of the parasite is restricted to blood, dual RNA-sequencing of host and parasite transcripts in blood can reveal their interactions at a systemic scale.

Using reference expression signatures for human leukocytes and parasite developmental stages we established that inter-individual heterogeneity in leukocyte and parasite populations explained a large proportion of the variation in gene expression between samples. After adjustments for leukocyte and parasite proportions, we identified human and parasite gene expression associated with severe disease features in Gambian children. Differences in parasite load explained up to 99% of differential expression of human genes but only a third of the differential expression of parasite genes. Co-expression analyses showed a remarkable co-regulation of host and parasite genes controlling translation, and host granulopoiesis genes uniquely co-regulated and differentially expressed in severe malaria. Our results indicate that high parasite load is the proximal stimulus for severe P. falciparum malaria, that there is an unappreciated role for many parasite genes in determining virulence, and hint at a molecular arms-race between host and parasite to synthesise protein products.

KEY WORDS

Malaria, RNA-seq, host-pathogen interactions

5:10 PM - 5:30 PM

National Centre for Indigenous Genomics

Invited Abstract

Dr Hardip Patel
National Centre for Indigenous Genomics, ANU

BIOGRAPHY:

Hardip is the bioinformatics lead for the National Centre for Indigenous Genomics.

ABSTRACT:

Hardip Patel[1], Yu Lin[2], Ashley Farlow[3], Tim McInerney[1], Renzo Balboa[1], Matthew Silcocks[3], Stephen Leslie[3], Simon Easteal[1]

[1] National Centre for Indigenous Genomics, The Australian National University, Acton, ACT - 2601
[2] Research School of Computer Science, The Australian National University, Acton, ACT - 2601
[3] Murdoch Children's Research Institute, Parkville Victoria - 3052

Genomics is rapidly progressing to enhance our understanding of functional role of the genome, genetic basis of disease and health and human history. However, it is not representative thus far. Initiatives such as the Korean Genome Project and H3 Africa have clearly indicated the importance of inclusion of diverse array of populations in genomic studies if we were to provide the benefits of genomics revolution equally to all. The National Centre for Indigenous Genomics (NCIG) was thus established in 2016 through the Federal Statute of the Australian National University Act 1991 to ensure that Indigenous Australians benefit equally and are at the forefront of the genomics revolution. NCIG, through Indigenous led governance board and state-of-the-art data repository, will provide the essential link between Indigenous and research communities that is needed for sustainable benefit and capability development. Towards those goals, ae at the NCIG have performed whole genome sequencing from 119 Indigenous Australians (~400 by the end of 2018) using Illumina platform at >30x coverage to establish a reference variation set. We have thus far discovered ~12 million SNPs (>25% novel) and ~12,000 structural variants with >1000 SNPs as high frequency variants. We have generated reference genomes using multitude of technologies (Illumina, PacBio, MinION, and Chromium 10X) from 4 individuals to identify the effects of largely European reference genome in identification and interpretation of variant calls in population specific studies. Our assembly of three genomes have revealed high degree of concordance with GRCh38. However, our main focus would be to identify and make use of novel and divergent sections of the genomes that have not been described yet for its utility in genomics study. This resource and enduring community engagement will ensure long term utility and benefits for all going forward.

KEY WORDS

Indigenous genomics, Whole genome sequencing, Reference genome assembly

SESSION 7: Plants

Chairs: Matthew Gilliham & Juan Carlos Sanchez Ferrero

9:00 AM – 9:45 AM

Simple model plant systems for whole organism engineering

Keynote Speaker
Jim Haseloff
University of Cambridge

BIOGRAPHY:

Jim Haseloff is the Head of the Haseloff lab, with a history of research in plant viroids, RNA enzymes and engineering approaches to plant development. Current interests are in simple open systems for plant synthetic biology including programmable cell-free extracts.

ABSTRACT:

Prof. Jim Haseloff

Department of Plant Sciences, University of Cambridge, Downing Street, Cambridge CB2 8AL UK.

Synthetic Biology is an emerging field that employs engineering principles for constructing genetic systems. It is providing a conceptual and practical framework for the systematic engineering of gene expression and cell behaviour in plants. We are using the liverwort Marchantia polymorpha as a simple plant system and testbed for bioengineering. Liverworts are characterised by morphological simplicity, matched by simple underlying genome structure. The ease of culture, transformation and analysis of Marchantia make it an ideal system for plant development and synthetic biology. We have developed a battery of computational, imaging and genetic tools to allow clear visualisation of individual cells inside living plant tissues, and developed a common syntax for plant DNA parts that can be used to reprogram metabolism and development.

The OpenPlant research initiative has created a hub for interdisciplinary exchange between the fundamental and applied sciences for plant agriculture and bioproduction. Its aim is to establish frameworks for the open exchange of new plant tools and DNA components that will promote innovation and international scientific exchange. OpenPlant brings together a wide range of engineers, scientists and policy developers to explore new technologies and possible models for sustainable agriculture, bioproduction and land use. Open tools provide an opportunity to democratise and improve international technology transfer, education and research training.

https://www.openplant.org
https://www.biomaker.org

9:45 AM - 10:10 AM

Investigating the Role of Structural Variation in Brassica Disease Resistance

Invited Speaker
Professor Jacqueline Batley
University of Western Australia, Crawley, Australia

BIOGRAPHY:

Prof Jacqueline Batley is an ARC Future Fellow at the University of Western Australia. She was awarded her PhD from the University of Bristol UK in 2001. She moved to Australia in 2002, as a senior research scientist at DPI-Victoria, then led a research group at the University of Queensland as an ARC QEII Research Fellow, from 2007-2014. Jacqui has received several awards for her research including a University of Queensland Foundation Research Excellence Award, an ARC QEII Fellowship and an ARC Future Fellowship. Jacqueline has expertise in the field of plant molecular biology, genetics and genomics, gained from working in both industry and academia. Her areas of interest include genetic and genomic analysis and specifically, genome sequence analysis, pan genomics, SNP analysis and the role of structural variation for applications such as genetic diversity, genetic mapping, LD, GWAS, evolutionary, population and comparative genomic studies, as well as the molecular characterisation of agronomic traits. She is currently focussing on blackleg resistance in Brassicas.

ABSTRACT:

Jacqueline Batley[1]

[1] School of Biological Sciences, University of Western Australia, Crawley, WA 6009

The Brassicaceae family contains some of the world's most important economic and agronomic crops, which are utilised as edible and industrial oilseeds (e.g Brassica napus, B. juncea) and vegetables (e.g Brassica oleracea, Raphanus raphanistrum), along with the scientific model plant Arabidopsis thaliana and highly diverse wild species. Pathogens, such as Leptosphaeria maculans (causal agent of Blackleg) and Sclerotinia sclerotiorum (causal agent of Sclerotinia stem rot), severely affect production of important crop species from the Brassicaceae. Plant genomes harbour resistance (R) genes, which play an important role in plant immunity, where nucleotide-binding site-leucine-rich repeat (NBS-LRR) genes are the most common type of R gene, followed by Receptor Like Proteins (RLPs) and Receptor Like Kinases (RLKs).

The identification of genes underlying important quantitative trait loci is extremely challenging in complex genomes such as B. napus. Recent advances in next-generation sequencing (NGS) has enabled development of millions of SNPs. However, as an increasing number of genome sequences become available, there is a growing understanding that the genome of a single individual is insufficient to represent the gene diversity within a whole species. We have examined the SNP diversity within R genes, and this allelic variation is an important source of phenotypic variation. However, we have also observed significant presence absence and copy number variation of R genes. We have developed Brassica pan genomes and using these the molecular analyses of candidate resistance genes using NGS data are presented. The difficulties associated with identifying functional gene copies within the highly duplicated Brassica genomes will be discussed. This analysis provides a valuable resource for the identification of R genes for enhanced crop protection through analysis of gene diversity and evolution linked to disease.

KEY WORDS

Resistance genes, pan genomics, presence absence variation, copy number variation

10:10 AM - 10:30 AM

Roles of long non-coding RNAs in Plant Sexual Reproduction

Invited Abstract

Dr Agnieszka Golicz
University of Melbourne

BIOGRAPHY:

Dr Agnieszka Golicz joined the University of Melbourne in July 2017 as a McKenzie Fellow working within the Plant Molecular Biology and Biotechnology Laboratory. She is a bioinformatician with interest in plant evolution and development. She has completed an undergraduate degree in Molecular Genetics from the University of Dundee in Scotland and a PhD in Applied Bioinformatics at the University of Queensland. Her graduate research focused on exploring the pangenomes and the genomic diversity within the representatives of the species Brassica oleracea. Currently she is focusing on integrating multi-omics datasets and using machine learning approaches for discovery and functional annotation of long non-coding RNAs. She is hoping to uncover long non-coding RNAs which are crucial for plant development and sexual reproduction, and contribute to a better understanding of their functions and mechanisms of action. She also enjoys exploring genomes of unusual plant species, for example seagrasses.

ABSTRACT:

Agnieszka A. Golicz[1], Prem L. Bhalla[1], Mohan B. Singh[1]

[1] Plant Molecular Biology and Biotechnology Laboratory, Faculty of Veterinary and Agricultural Sciences, University of Melbourne, Parkville, Melbourne, VIC, Australia

Long non-coding RNAs (lncRNAs) are transcripts over 200 base pairs in length with no discernible protein coding potential. Recently lncRNAs have emerged as important stage specific regulators of protein gene expression in both plants and animals. Studies in plants and animals consistently show high expression of lncRNAs in male reproductive organs in cell and tissue-specific manner. Analogous to animal systems, in most land plants sexual reproduction requires pollination involving transfer of the male sperm carried in the pollen to the female part of a flower, the stigma. In rice, isolation of individual pollen components including the sperm cell and the supporting vegetative cells has become possible allowing study of their transcriptomes. We have performed rice genome re-annotation and lncRNA discovery and studied lncRNA expression across transcriptomes of a diverse panel of rice tissues and cell types, including the sperm cells. We show that protein coding and lncRNA genes have divergent expression profiles and that the rice sperm cells express more lncRNAs than any other tissue/cell type. We further describe a method developed to characterize the lncRNAs using heterogenous evidence including genomic, transcriptomic and phenotypic data. Our results suggest that high expression of lncRNAs in male-specific tissues is a developmental hallmark of both animals and plants and likely a result of convergent evolution. We hypothesize the roles of lncRNAs as master regulators of gene expression and chromatin organization might make them particularly suited for coordination and control of molecular processes involved in sexual reproduction.

KEY WORDS

Plant sexual reproduction, male fertility, long non-coding RNAs, rice, Oryza sativa

11:00 AM - 11:25 AM

Genome Sequencing, Assembly and Annotation of the Industrially Useful Plant Plantago ovata

Invited Speaker
Professor Rachel Burton
University of Adelaide

BIOGRAPHY:

Professor Rachel Burton is based at the Waite Campus, University of Adelaide. She is a plant scientist and molecular biologist, passionate about plant cell walls and all the useful things they can do for us. She is interested in the ways that the cell wall components are made and put together but even more intrigued by how they are disassembled or fermented in the human gut, because they are the crucial dietary fibre part of our diets. Rachel is also interested in renewable biofuels and believes that different plant feedstocks are fundamentally important to meeting our future energy needs.

ABSTRACT:

Julian Schwerdt[1], Neil J Shirley[1], Kylie Neumann[1], Matthew R Tucker[1] and **Rachel A Burton[1]**

[1] ARC Centre of Excellence in Plant Cell Walls, School of Agriculture, Food and Wine, University of Adelaide, Urrbrae, SA 5064

Plantago species are myxospermous; their seeds extrude a mucilage when they encounter an aqueous environment, aiding seed dispersal and germination. The mucilage of some Plantago species have been used for hundreds of years as herbal remedies and in modern times we are most familiar with psyllium which comes from Plantago ovata. Psyllium is the papery husk milled off the outside of the dry mature seed. This husk predominantly contains heteroxylan, a branched polysaccharide with significant water-holding capacity that is valuable as a dietary fibre supplement providing bulk and aiding laxation, but also acts as a versatile hydrocolloid in processed food.

As well as use in products such as ice cream and as a key ingredient in gluten-free foods, psyllium is also finding environmental applications in green concrete and as a corrosion inhibitor. Here we describe the ~582Mb Plantago ovata draft genome assembly of ~70X coverage PacBio reads. The canu assembly is composed of 7181 contigs with an NG50 of 227945. First-round automatic annotation predicted139614 transcripts and was performed using the BRAKER2 pipeline; with evidence sets from nine RNAseq libraries comprising whole leaf, leaf with no vascular bundle, bract and internode tissues. HiC scaffolding is currently being attempted and will be followed by second-round annotation and functional characterisation. As Plantago mucilage is primarily composed of heteroxylans a draft genome sequence provides the opportunity to characterise genetic and biochemical pathways associated with heteroxylan biosynthesis. We observed expansion and diversification of xylan-associated enzymes including genes from the Glycosyltransferase 61 family. These enzymes have been associated with the substitution of the xylan backbone and have been shown to co-vary with seed coat mucilage composition across Plantago species.

KEY WORDS

PacBio, canu, seed mucilage, psyllium

11:25 AM - 11:45 AM

Population sequencing reveals clonal diversity and ancestral inbreeding in the grapevine cultivar Chardonnay

Invited Abstract

Dr Michael Roach
Australian Wine Research Institute

BIOGRAPHY:

Michael Roach is a Bioinformatics Post-Doctoral Researcher at the Australian Wine Research Institute. His work mainly focuses on genomics of wine-related organisms, but occasionally includes modelling of proteins and protein-ligand interactions. He is the creator and maintainer of Purge Haplotigs--a pipeline for curating third-gen genome assemblies. His is currently working on genomics of spoilage microorganisms, as well as unravelling the heritage of some of the oldest grapevine cultivars.

ABSTRACT:

Michael J. Roach[1], Daniel L. Johnson[1], Joerg Bohlmann[2], Hennie J.J. van Vuuren[2,3], Steven J. M. Jones[4], Isak S. Pretorius[5], Simon A. Schmidt[1]* and Anthony R. Borneman[1,6]*

[1] The Australian Wine Research Institute, PO Box 197, Glen Osmond, South Australia, 5046, Australia
[2] Michael Smith Laboratories, The University of British Columbia, Vancouver, British Columbia, Canada
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[4] Michael Smith Genome Sciences Centre, British Columbia Cancer Research Centre, Vancouver, British Columbia, BC V6T 1Z4, Canada.
[5] Chancellery, Macquarie University, Sydney, New South Wales, 2109. Australia
[6] Department of Genetics and Evolution, University of Adelaide, South Australia, 5000, Australia
* Authors contributed equally to this work

Chardonnay is the basis of some of the world's most iconic wines and its success is underpinned by a historic program of clonal selection. There are now numerous clones of Chardonnay available that exhibit differences in key viticultural and oenological traits that have arisen during the long-term asexual propagation of this woody-plant species. However, the genetic variation that underlies these differences remains largely unknown.

A highly contiguous diploid-phased Chardonnay genome assembly was produced, and combined with re-sequencing data from 15 different commercial Chardonnay clones. We identified 1620 markers that distinguish the 15 Chardonnay clones. These were reliably used in clonal identification from independently-sourced and sequenced material. Furthermore, we identified many marker variants that may account for phenotypic differences between clones, including the known and well-characterised 'Muscat' mutation.

We used sequencing data from the parents of Chardonnay—Gouais blanc and Pinot noir—to map the inheritance over the entire Chardonnay genome. We detected instances of heterosis, with differentially-expanded gene families inherited from the parents of Chardonnay. Most surprisingly, the patterns of nucleotide variation present in the Chardonnay genome indicate that the parents of Chardonnay share an extremely high degree of kinship.

A high-quality, diploid-phased Chardonnay assembly has greatly furthered understanding the genetics and heritage of this important cultivar. We demonstrate a reliable method to identify unknown grapevines to a specific clone. Finally, we show evidence of a more complicated pedigree for Chardonnay than previously reported, with Gouais blanc and Pinot noir possibly sharing an immediate-family relationship.

KEY WORDS

clonal variation; genetic variability; Vitis vinifera; PacBio SMRT long-read sequencing; parentage assignment; kmer genotyping

11:45 AM - 12:05 PM

Functional Structure and Association of Root Microbiome with Foxtail Millet Yield Unravelled by Ultra-Deep Metagenomic Sequencing

Invited Abstract

Dr Yayu Wang
BGI Shenzhen

BIOGRAPHY:

Dr. Yayu Wang completed her degrees in plant genetics. She has more than 9 years' experience in environmental microbial research and is familiar with microbial genomics, metagenomics and metatranscriptomics approaches and applications. Dr. Wang and her current team are working on scientific research of plant root microbial ecosystems, focusing on functional adaption and evolutional dynamics of the root microbiome from grain crops, vegetables, fruit trees and special plants. Their primary interest is in using multi-omics technologies to unravel the molecular basis by which host plant, microbes and environmental variables interactions and then model their interactions to assist plant growth. She has published 4 research papers in the related topics and is involved in one Chinese government supported scientific research project.

ABSTRACT:

Yayu Wang[1,2], Pengfan Zhang[1,2], Huan Liu[1,2], Xin Liu[1,2]

[1] BGI-Shenzhen, Shenzhen 518083, China
[2] China National GeneBank, BGI-Shenzhen, Shenzhen 518120, China

Plant roots assemble a subset of soil-borne microbiota which can regulate plant growth and health. Amplicon sequencing has revealed the taxonomic structure of many plant root microbiomes. However, little is known about the functional structure of root microbiome and the association between microbial functional genes with plant traits. Here, we used foxtail millet (Setaria italica), a traditional crop plant of China as a model to study the root microbiome functional composition and its association with plant yield by ultra-deep coverages. We established a comprehensive reference gene catalog for foxtail millet root-associated microbiome using more than 2 tera bases (TB) metagenome data by a rapid and novel protein-based method. We also evaluated the required metagenomic sequencing depth for rhizosphere microbiome (46 giga bases (Gb)). Further, we found compartment-specific enriched metabolic pathways and carbohydrate-active enzymes (cazymes) by comparing functional traits between rhizosphere and rhizoplane microbiomes. Finally, we identified 38 KEGG Orthology (KOs) and 9 Metagenomic Species (MGS) which were positively correlated with foxtail millet yield. Our work provides the foundation for root-associated metagenomic study, and further advances our understanding of root-associated microbiome. Particularly, the potential functional biomarkers may be anticipated to further improve the foxtail millet yield and augment its sustainable cultivation.

KEY WORDS

Root microbiome, Setaria italica, metagenome, productivity

SESSION 8: Metagenomics

Chairs: Renee Smith & Anthony Borneman

1:05 PM - 1:50 PM

Mosaic of microbes: Investigating the involvement of marine microbiomes in the life and death of kelp forests

Keynote Speaker
Dr Elizabeth Dinsdale
San Diego State University

BIOGRAPHY:

Dr. Dinsdale's lab at San Diego State University uses next-generation sequencing to tease apart ecological and evolutionary relationships within marine, coral reef and kelp forest ecosystems. We are a multi-disciplinary team of students and researchers from around the globe.

Dr. Dinsdale helped establish one of the first university-level classes to offer hands-on experience with a pyrosequencing machine. In 2011, students of BIOL596 Ecological Metagenomics at SDSU sequenced the California sea lion genome at 12X coverage. Students applied bioinformatics and statistics techniques to analyze the resulting genomes and metagenomes.

ABSTRACT:

Megan M Morris[1], Michael P. Doane[1], J. Matthew Haggerty[2], Bhavya Papudeshi[3], Lais Lima[1], Dnyanada Pande[4], Robert. A Edwards[1], **Elizabeth A Dinsdale[1]**

[1] San Diego State University, San Diego CA USA
[2] University of Southern California, Los Angeles, CA USA
[3] National Center for Genome Analysis Support, Indiana University, Bloomington, Indiana, USA
[4] Fred Henry Cancer Center, Settle, USA

Marine microbes are diverse, numerous, and we use random shot-gun metagenomics, bioinformatics and a range of statistical analysis to investigate how microbiomes affect the ecology of an iconic marine macro-alga; Macrocystis pyrifera. From a study starting in 2010, we show that water column and kelp frond associated microbes are distinctive, and the water column microbiomes are influenced by abiotic processes, while the kelp associate microbiomes are not. The kelp microbiomes remain relatively stable over the length of the 30 m plant, but showed fluctuations in late summer, associated with kelp degradation. As the kelp degrades, the microbiome undergoes a successional change from ~ 25 % Pseudomonas to ~ 35 % Pseudoaltermonas and dominance by Sulfidobacter (~ 60 %) at late stages of degradation. The microbial succession occurred in all 12 experimental units, under both 12 and 20oC temperature conditions. A binning approach identified novel population genomes, however, some over-represented genes in the microbiome were within the rare biosphere. As kelp grow, they bioaccumulate heavy metals, such as copper, particularly near human activity, causing the microbes to have more heavy metal resistance genes. These genetic changes in the microbiome caused a decline in kelp recruitment and settlement. During our sampling period, a non-native species, Sargassum hornei invaded the kelp forest affecting the microbes on the kelp and in the surrounding environment. The S. horneri had distinctive microbiomes that become dominated by Vibrio species over time. The Vibrio species started to leach into the benthic biofilms, and we predict this will also affect kelp recruits. The microbiome of the kelp growing near the S. horneri showed dysbiosis, and the kelp showed signs of bleaching, suggesting that microbial warfare is used during the invasion process. NSG has enabled us to determine microbes effect recruitment, growth, and degradation of kelp.

KEY WORDS

Metagenomics, kelp forests, population genomes, bioninformaitc

1:50 PM - 2:15 PM

A TCE contaminated aquifer reveals a Piggyback-the-Persistent viral strategy

Invited Speaker
Dr James Paterson
Flinders University

BIOGRAPHY:

I am currently a Postdoctoral Research Fellow in the Microbial Systems Laboratory under the supervision of Prof. Jim Mitchell. My main area of research is focused on microbial community composition and dynamics within aquifer systems and how this information can define aquifer ecosystem state.

ABSTRACT:

James S. Paterson[1], Renee J. Smith[1,2], Jody C. McKerral[1], Lisa M. Dann[1], Elise Launer[1], Peter Goonan[3], Tavis Kleinig[3], Jed A. Fuhrman[4] and James G. Mitchell[1]

[1] College of Science and Engineering, Flinders University, GPO Box 2100, Adelaide, SA 5001, Australia
[2] College of Medicine and Public Health, Flinders University, GPO Box 2100, Adelaide SA 5001, Australia
[3] South Australia Environment Protection Authority, GPO Box 2607 Adelaide SA 5001, Australia
[4] Department of Biological Sciences, University of Southern California, 3616 Trousdale Parkway, Los Angeles, CA 90089, USA

Subsurface environments hold the largest reservoir of microbes in the biosphere. They play essential roles in transforming nutrients, degrading contaminants and recycling organic matter. The global demand for freshwater makes aquifers important water sources, and contaminated sources are being used increasingly. Here, we propose a previously unrecognized fundamental microbial process that influences aquifer bioremediation dynamics and that applies to all microbial communities. In contrast to previous models, our proposed Piggyback-the-Persistent (PtP) mechanism occurs when viruses become more dominated by those exhibiting temperate rather than lytic lifestyles driven by persistent chemicals (in our case pollutants) that provide long term carbon sources and that refocus the aquifer carbon cycle, thus altering the microbial community. Our model system is a chlorinated-hydrocarbon contaminated aquifer. The contaminant of the aquifer is slow to degrade and has kept bacterial abundance relatively low. In this ultra-oligotrophic system, the virus:microbial ratio (VMR) ranges from below the detection limit of 0.0001 to 0.6, well below the common aquatic range of 3-10. Shortest-average-path network analysis revealed VMR and trichlorethene (TCE) as nodes through which ecosystem information and biomass most efficiently pass. Novel network rearrangement revealed a hierarchy of Kill-the-Winner (KtW), Piggyback-the-Winner (PtW) and PtP nodes. We propose that KtW, PtW and PtP occur simultaneously as competing strategies, with their relative importance depending on conditions at a particular time and location. Unusual nutrient sources, such as TCE, appear to contribute to a shift in this balance, which in turn provides a mechanism for quantifying contamination impact in microbially dominated food webs.

KEY WORDS

Microbial ecology, 16S rRNA, flow cytometry

2:15 PM - 2:40 PM

Tracking evolution in experimental populations of microbes using time-resolved whole genome sequencing.

Invited Speaker
Dr Michael McDonald
Monash University

BIOGRAPHY:

Mike completed his Bachelor and Master's degrees (both in Genetics) at Otago University in New Zealand. Mike started his PhD at Auckland and finished it at Massey University where he used P. fluorescens as an experimental model of evolutionary genetics. Mike's first Postdoc was at Academia Sinica in Taiwan, where he learned Yeast genetics and a little Chinese. Mikes second Postdoc was at Harvard University at the FAS Center for Systems Biology, where he used whole genome sequencing and high-throughput methods to study evolution in Yeast and E. coli. Mike is currently a ARC Future Fellow and Group Leader in the School of Biological Sciences at Monash University.

ABSTRACT:

Michael McDonald[1]

1 Monash University, VIC Australia

Experimental evolution is a powerful method for testing fundamental questions in evolution and ecology. I will present recent work showing how high-throughput sequencing methods can provide insights into a classic problem in evolutionary biology, the evolution of sex, as well as eco-evolutionary dynamics in experiment populations of E. coli (the Lenksi LTE) and and yeast. Currently, our understanding of the evolution and ecology of natural communities comes from the "top down" approaches of community ecology and metagenome sequencing. Although experimental evolution has the potential to contribute, most microbial evolution experiments are in laboratory settings far removed from the actual conditions that microbes in the wild would experience. My work seeks to connect these two fields, so that the mechanistic insights possible in the lab are applied in experimental settings that better approximate natural and clinical environments.

2:40 PM - 3:00 PM

A genomic autopsy of perinatal death

Invited Abstract
Miss Alicia Byrne
Centre for Cancer Biology

BIOGRAPHY:

Alicia Byrne is a graduate student, in the final year of her PhD at the University of South Australia. Her research investigates the utility of a 'genomic autopsy', using whole exome or whole genome sequencing, to better identify the causes underlying stillbirth and newborn death.

ABSTRACT:

Byrne A[1,3,9], Weyrich L[2], Eisenhofer R[2], Walker M[3], Nguyen H[4], Vlamakis H[3], Schreiber A[5,9,10], Feng J[5,9], Soubrier J[4], Babic M[1], Khong Y[6], Moore L[6,11], MacArthur D[3], Hahn C[1,9,11], Kassahn K[4,10], King-Smith S[1,7], Barnett C[8,11], Scott H[1,4,5,7,9-11]

1 Molecular Pathology Research Laboratory, Centre for Cancer Biology, Adelaide, SA, Australia
2 Australian Centre for Ancient DNA, University of Adelaide, Adelaide, SA, Australia
3 Broad Institute of MIT and Harvard, Boston, MA, USA
4 Department of Genetics and Molecular Pathology, SA Pathology, Adelaide, SA, Australia
5 ACRF Cancer Genomics Facility, Centre for Cancer Biology, Adelaide, SA, Australia
6 Department of Surgical Pathology, Women's and Children's Hospital/SA Pathology, North Adelaide, SA, Australia
7 Australian Genomics Health Alliance, Melbourne, VIC, Australia
8 SA Clinical Genetics Service, Women's and Children's Hospital/SA Pathology, North Adelaide, SA, Australia
9 School of Pharmacy and Medical Sciences, University of South Australia, Adelaide, SA, Australia
10 School of Biological Sciences, University of Adelaide, Adelaide, SA, Australia
11 School of Medicine, University of Adelaide, Adelaide, SA, Australia

Pregnancy loss or newborn death are devastating events, and precise identification of the cause is essential for accurate counselling. Congenital abnormalities are the leading cause of death (33%), followed by infection (15%), however, the etiology of malformations is not always apparent and a causative agent cannot always be determined in cases with infectious pathology. Here, we examine the utility of a 'genomic autopsy' in identifying microbial species causative of fetal death. Whole genome sequencing is being performed on DNA extracted from fetal lung, with different protocols trialled to optimise dual extraction of human and microbial DNA. Sequence reads are mapped to a set of microbial reference genomes and a metagenomics approach employed to determine microbial load and identify causative organisms.
A proof-of-principal study has been performed on 20 cases, 10 due to infection (positive) and 10 due to congenital abnormality without a molecular cause (negative). Causative microbes were correctly identified in 4/7 known positive cases and plausible microbes identified for 2/3 unsolved positive cases. Methods are now being optimised to improve diagnostic yield. As expected, no significant microbial load was identified in 9/10 negative cases, however, a known pathogen was identified in 1 case. Analysis of human-mapping reads also revealed a genetic variant in 1 positive case, upon review the patient's phenotype shared similarities to that expected for the gene. These cases demonstrate the phenotypic overlap that can occur in congenital disorders with infectious or genetic origin, and highlights the need for a testing approach that can simultaneously assess both.
Together, the dual analysis of human and microbial DNA that a genomic autopsy allows may facilitate an accurate cause

of death to be established in up to 50% of cases.

KEY WORDS
Perinatal, Metagenomics, Diagnostics

3:00 PM - 3:20 PM

Genome based metagenomic analysis using extensive human gastrointestinal culture collection

Invited Abstract

Dr Samuel Forster
Hudson Insitute of Medical Research

BIOGRAPHY:

Dr Samuel Forster has recently established the Microbiota and Systems Biology Laboratory within the Centre of Innate Immunity and Infectious Diseases at the Hudson Institute of Medical Research. Previously at the Sanger Institute, Sam's research resulted in the development of methods to culture the majority of human gastrointestinal microbiota, advanced algorithms for metagenomics analysis and identification of rationally selected bacteriotherapy candidates. Research within the Microbiota and Systems Biology laboratory combines microbiology, immunology and computational biology to understand the functional role of the microbiota and determine ways to modify these communities to improve human health.

ABSTRACT:

Samuel C Forster[1,2], Junyan Liu[2], Nitin Kumar[2] and Trevor Lawley[2]

[1] Microbiota and Systems Biology Laboratory, Centre for Innate Immunity and Infectious Diseases, Hudson Institute of Medical Research, Clayton, Victoria, Australia, 3168
[2] Host-Microbiota Interactions Laboratory, Wellcome Sanger Institute, Wellcome Genome Campus, United Kingdom, CB10 1SA

The human gastrointestinal microbiota represents a new frontier in our understanding of human biology; however, our current knowledge is hampered by the limited number of purified bacterial isolates and the lack of reference genomes to enable detection and guide functional analysis. We have recently generated the Human Gastrointestinal Bacteria Culture Collection (HBC), a comprehensive archive of 737 whole-genome sequenced bacterial

isolates, representing 273 species (105 novel species) from 31 families found within the human gastrointestinal microbiota. This collection enables classification of 83% of genera by abundance across 13,490 shotgun sequenced metagenomic samples, improves taxonomic classification by 61.0% compared to the Human Microbiome Project (HMP) genome collection alone. Applying this resource, we have developed methods to identify and track movement of AMR containing mobile elements between key gastrointestinal pathogens and the commensal microbiota and have validated these computational predictions using conventional conjugation assays.

KEY WORDS

Microbiota, metagenomics, gastrointestinal, computational biology, culturing, antimicrobial resistance

3:20 PM - 3:45 PM

Expanding our view of microbial communities in thawing permafrost

Invited Speaker
Gene Tyson
The University of Queensland

ABSTRACT:

Gene W. Tyson

Australian Centre for Ecogenomics, School of Chemistry & Molecular Biosciences, The University of Queensland, St Lucia, Queensland, Australia.

Over the last decade, metagenomics has changed the face of microbial ecology. Metagenomics bypasses traditional culture-dependent approaches and holds the promise of genome-level insights into the mostly uncharted microbial world. When combined with techniques for exploring gene/protein expression and metabolic activity we are able to explore the communities, interactions and processes that drive important ecosystems.

As global temperatures rise, large amounts of carbon sequestered in permafrost are becoming available for microbial degradation. Accurate prediction of carbon gas emissions from thawing permafrost is limited by our understanding of these microbial communities. To investigate these communities, metagenomic sequencing of 214 samples from a permafrost thaw gradient was used to recover 1,529 metagenome-assembled genomes, including many from phyla with poor genomic representation. These genomes reflect the diversity of this complex ecosystem, with genus-level representatives for more >60% of the community. Meta-omic analysis revealed key populations involved in the degradation of organic matter, including bacteria whose genomes encode a previously undescribed fungal pathway for xylose degradation. Microbial and geochemical data highlight lineages that correlate with the production of greenhouse gases and indicate novel syntrophic relationships. Our findings link changing biogeochemistry to specific microbial lineages involved in carbon processing, and provide key information for predicting the effects of climate change on permafrost systems.

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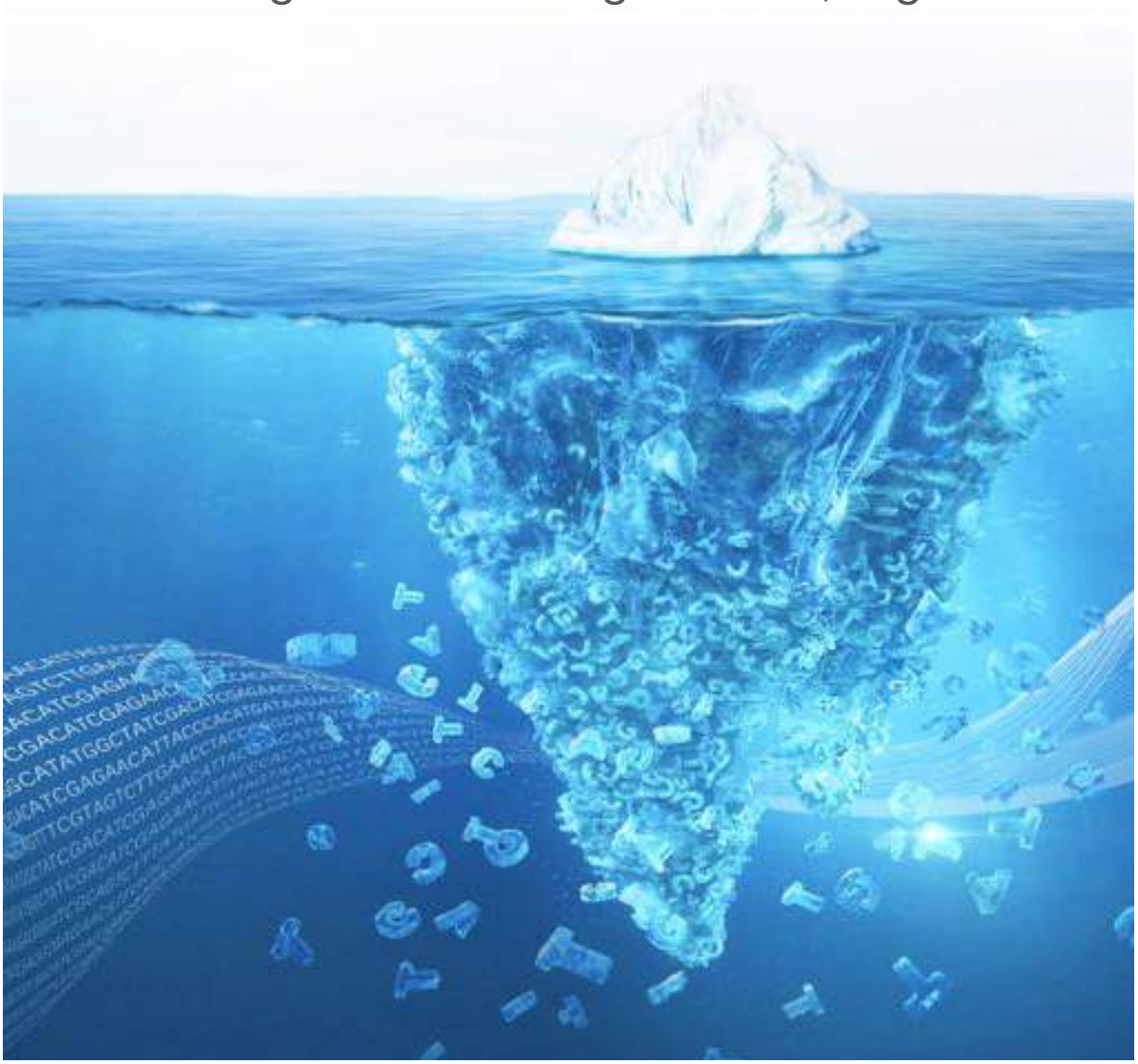

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POSTER SESSIONS

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#	POSTER TITLE	PRESENTER
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3	Detection of dicistronic tRNA:mRNA transcripts in grapevine (Vitis vinifera)	Na Sai
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POSTER 1

Validation of the ForenSeq™ DNA signature prep kit and the use of ancestry and phenotype informative markers for forensic applications

Mr Ryan England
Institute of Environmental Science and Research (ESR), Auckland, New Zealand

BIOGRAPHY:

Ryan England is a Research Scientist at ESR in the Forensic Research & Development team. In 2017 Ryan began a PhD with the University of Auckland. He works on the Forensic Genomics project, which aims to assess and validate the use of massively parallel sequencing for sequencing forensic DNA samples. The project focuses on sequencing markers such as STRs, ancestry and phenotype informative SNPs and the human mitochondrial genome. Ryan is also the sequencing facility coordinator at ESR's Auckland site, where he coordinates the sequencing of samples for multiple research projects. He is experienced in MPS sample and library preparation, sequencing and bioinformatics. Ryan joined ESR in 2013 after completing his MSc at Massey University, where he used MPS to sequence New Zealand native bird mitochondrial genomes.

ABSTRACT:

Ryan England[1,2], Janet Stacey[1], Gemma Nancollis[1], Andrew Sarman[1] and SallyAnn Harbison[1]

[1] Forensic Business Group, Institute of Environmental Science and Research (ESR), Private Bag 92021, Auckland 1025, New Zealand
[2] School of Chemical Sciences, The University of Auckland, Private Bag 92019, Auckland, New Zealand

Forensic biology is currently undergoing a transformation towards the use of massively parallel sequencing. At ESR we are validating the ForenSeq™ DNA Signature Prep Kit and the MiSeq FGx™ Sequencer from Verogen© according to the SWGDAM guidelines for forensic validation.

The ForenSeq™ DNA Signature Prep Kit amplifies both Short Tandem Repeats (STRs) and SNPs all in a single multiplex reaction, enabling back compatibility with current STR capillary electrophoresis data, as well as sequencing ancestry and phenotype informative SNPs. These ancestry and phenotype informative SNPs can be used to predict what someone looks like to provide intelligence information for a criminal investigation.

This presentation will describe the work we have done to evaluate the performance of the ForenSeq™ DNA Signature Prep Kit, including modifications we have made to optimize sequencing performance. We have studied how low level DNA samples and casework type samples effect the sequencing results and address technical issues that require consideration when sequencing DNA below the optimum amounts recommended. We demonstrate that informative DNA sequence profiles can be obtained from very small numbers of laser micro-dissected epithelial and sperm cells, including those from cell mixtures. We also present how the sequencing data is processed and analysed, in particular the ancestry and phenotype informative markers sequencing data. We look at what tools are available to use this SNP data to predict what an individual looks like and how this information could be useful to a criminal investigation.

KEY WORDS

Forensic biology, massively parallel sequencing, Short Tandem Repeats, ancestry informative, phenotype informative

POSTER 2

Viking horses of the North Atlantic

Dr Heidi Nistelberger
University of Oslo, Oslo, Norway

BIOGRAPHY:

Heidi Nistelberger is a postdoctoral research fellow at the University of Oslo, researching the impact of Vikings on the genomic composition of important domesticated species including horses.

ABSTRACT:

Heidi Nistelberger[1], Bastiaan Star[1], Albina Pálsdóttir[1], Agata Gondek[1], Rúnar Leifsson[2], Jón Hallsson[3], Jan Bill[1], Emma Svensson[4], Anne Karin Hufthammer[5], James Barrett[6], Ludovic Orlando[6], Sanne Boessenkool[1]

[1] University of Oslo
[2] The Cultural Heritage Agency of Iceland
[3] The Agricultural University of Iceland
[4] Uppsala University
[5] University of Bergen
[6] University of Cambridge

Horses were of central importance to Viking culture and were utilised for transport, warfare and agricultural purposes. They were also status symbols and the most common grave good associated with Viking Age burials in Iceland. Using shotgun sequencing and targeted enrichment of ancient DNA, we investigated the genomic composition of the largest collection of Viking Age horses from the North Atlantic to date, with an aim to reveal the sex, phenotypic traits and demographic patterns associated with these animals. Our findings will deepen our understanding of the use and status of horses during the Viking Age.

POSTER 3

Single Cell Whole Genome Sequencing – A Comparison of NGS Platforms

Dr Allan Motyer
University of Melbourne, Parkville, Australia

BIOGRAPHY:

Dr Allan Motyer is a statistical geneticist whose research has focused on statistical methods for the interrogation of immune-system genes and their analysis in genetic association studies. His current work involves the bioinformatic and statistical analysis of next-generation sequencing data, specifically single-cell whole genome sequencing of multiple sclerosis patients. Dr Motyer obtained his PhD in mathematics from the University of Melbourne in 2011. He is currently a postdoctoral researcher in the statistical genetics group of Associate Professor Stephen Leslie. Dr Motyer was awarded the prize for best early career researcher presentation at the Australian GeneMappers 2017 conference.

ABSTRACT:

Allan Motyer[1], Stacey Jackson[2], Antony Harding[3], Michael Barnett[3] Lynn Fink[4], Stephen Leslie[1], Justin Rubio[2,5]

[1] Melbourne Integrative Genomics, Schools of Mathematics and Statistics, and BioSciences, The University of Melbourne
[2] Department of Pharmacology and Therapeutics, The University of Melbourne
[3] Brain & Mind Research Institute, University of Sydney
[4] BGI Australia
[5] Florey Institute of Neuroscience and Mental Health

Single cell whole genome sequencing (scWGS) has the potential to reveal novel biological insights "hidden" from studies conducted on DNA from bulk tissue and/or mixed cell populations. By comparing the genomes of different single cells with matched 'bulk' DNA, somatic variation can be identified and used to improve our understanding of cell lineage development, mutational processes in normal cells and the role of somatic mutation in many diseases.

However, a single human nucleus contains only 5 pg of DNA, so a whole genome amplification (WGA) step is first necessary to generate a sufficient amount for sequencing. This WGA step introduces biases, such as allelic dropout, which can impact data fidelity and generate specific bioinformatic challenges to the acquisition of reliable variant callsets, and thus new platforms require robust validation prior to their adoption. To date, published scWGS data have been generated using Illumina short-read sequencing. In principle, other sequencing platforms can be used, but benchmarking quality metrics between platforms is necessary. Here, we have assessed the suitability of the BGI-SEQ platform for scWGS by comparison with Illumina NovaSeq. Ten single cell WGA samples (single neuronal nuclei isolated by FACS from frozen post-mortem neural tissue) and matched bulk DNA extracted from the brains of four individuals who had multiple sclerosis were sequenced to a depth of 45x on both platforms. We assessed various quality metrics such as read error rate and coverage, and the concordance of single nucleotide variant calls using several state-of-the-art variant callers. To our knowledge, this is the first time that the performance of Illumina and BGI-SEQ WGS platforms have been compared for sc DNA samples, so the results will allow researchers to make an informed decision about the platform most suitable for their scWGS study.

KEY WORDS

Single cell whole genome sequencing; NGS; BGI-SEQ; somatic mutation; bioinformatics

POSTER 4

DNA Methylation Wide Association analysis in Amyotrophic Lateral Sclerosis

Mrs Tian Lin
University of Queensland, Brisbane, Australia

BIOGRAPHY:

Tian Lin completed her PhD in Plant Biology in 2014. She has been working as a research assistant within the Program of Complex Trait Genomics within the Institute of Molecular Bioscience at the University of Queensland.

ABSTRACT:

Tian Lin[1] representing the sporadic ALS Australia Systems Genomics Consortium

[1] Institute for Molecular Bioscience, The University of Queensland, Brisbane, Queensland 4072, Australia

Amyotrophic lateral sclerosis (ALS), or Motor neurone disease (MND), is a fatal neurodegenerative disease with lifetime risk of ~0.3%. While about 5-10% of cases come from families with many affected members, the vast majority have no known family history. This so-called sporadic ALS is likely a complex genetic disease with both genetic and nongenetic risk factors. Here, we conduct a DNA-methylation-wide association study (MWAS) of 782 cases and 614 controls from Australia, comprised of two data cohorts. DNA methylation (DNAm) is measured in whole blood, assessed with the Illumina HumanMethylation450 array. DNAm differences between cases and controls may reflect both consequence and cause of disease. While differences may need careful interpretation, the MWAS approach can leverage opportunities to understand factors that contribute to disease onset and progress. We found that blood cell composition proportions predicted from DNAm were significantly different between ALS cases and controls, replicated across the cohorts.

In a mixed linear model association analysis, using our software, OSCA (omicS-data-based Complex trait Analysis), which fits the genome-wide epigenetic value of each person as random effect, we identified two probes that show significantly different DNA methylation between cases and controls. We used EWAS results from one cohort used the probe effect estimates to calculate a methylation score in the other cohort. This out-of-sample prediction ranked cases higher than controls with probability 0.66 (i.e. AUC=0.66), providing good evidence for significant DNAm differences attributed to ALS. Ongoing bioinformatic analyses are integrating our results with functional annotations.

POSTER 5

Bioinformatic challenges in the analysis of CLIP Experiments.

Dr Emily Hackett-Jones
Centre for Cancer Biology, University of South Australia

BIOGRAPHY:

Emily received her MSc at the University of Adelaide in subatomic physics, before moving to the UK to complete a PhD in theoretical and mathematical physics.

Emily's career has spanned both industry and academia, and a variety of different topics. After her studies in string theory, she returned to Australia in 2009 to begin research into biological systems and cell motion. She then spent five years working for Accenture as a data scientist, working in digital marketing, looking at how machine learning can aid business decisions.

Today Emily works as a Bioinformatician in the Centre for Cancer Biology at the University of South Australia. She has a particular interest in cancer metastasis, non-coding RNAs and analysis of HITSCLIP experiments. As a regular speaker for ChooseMaths and HerTechPath, she actively encourages women to pursue STEM careers.

ABSTRACT:

Hackett-Jones, E.J.[1,2], Pillman, K.A.[1,2], Toubia, J.[1,2], Dredge, K.[1], Bert, A.G.[1], Bracken, C.P.[1,2], Gregory, P.A.[1,2] and Goodall, G.J.[1,2]

[1] Centre for Cancer Biology, University of South Australia and SA Pathology, Australia
[2] University of Adelaide, Australia

MicroRNAs (miRs) are small non-coding RNAs known to bind - often via complementary seed sequences - to messenger RNAs and act repressively on cellular protein levels. Much of the interest in miRs concerns their role in cancer, as dysregulation of miRs is common in cancer. Although miR:mRNA binding sites can be predicted in silico, many of the millions of predicted sites are spurious. CLIP-Seq experiments involve high-throughput next generation sequencing of RNA cross-linked to miRs, and are a relatively new method to determine functional bindings of miRs to mRNAs.

We shall outline our development of a bioinformatic analysis pipeline for CLIP-Seq data, including peak calling, identification of PCR duplicates, and downstream motif analysis. We will discuss ways to improve the identification of predicted functional miR:mRNA binding sites, and recent experimental results.

KEY WORDS

bioinformatics, microRNA, RNA-seq, HITS-CLIP, cancer.

POSTER 6

Self-Terminated Random-Forest for Whole Genome GWAS analysis

Dr Natalie Twine
CSIRO, Australia

BIOGRAPHY:

Arash Bayat is a PhD student at School of Computer Science and Engineering of University of New South Wales (UNSW). He joins Transformational Bioinformatic Team of CSIRO as Postdoctoral fellowship. His research interest are algorithms and bioinformatics.

ABSTRACT:

Arash Bayat[1], **Natalie Twine[2]**, Laurence Wilson[1], Aidan R. O'Brien[1], Piotr Szul[2], Robert Dunne[2], Denis C. Bauer[1]

[1] Transformational Bioinformatics Team CSIRO
[2] Data61 CSIRO

Random-Forest is an ensemble machine learning method that aggregates the outcome of several decision trees. Random-Forest is particularly used in GWAS application due to its capability to consider interacting features. Random-Forest training is a progressive process in which the accuracy of the model is improved as more trees are built. Yet the improvement gained is decreased with the growth in the number of trees. The model can be considered stable when adding more trees does not improve the accuracy substantially.

For small-sized datasets, a large number of trees can be built to make sure that the model is stable. However, for larger datasets, the computational cost quickly becomes a consideration. For example, even for VariantSpark, a fast and parallelised implementation of Random-Forest on distributed computing architecture (Spark), it already takes 870 CPU hours to build 10 thousand trees for a moderate dataset of ~7.2 million features and ~2,000 samples.

There have been several efforts in the literature to identify the optimal number of trees to avoid over-computing. Here we propose a novel approach that captures the variation in the out-of-bag error and the variable importance during the training process. When the variation converges, the model is considered stable and the training process is terminated.

Considering both the prediction accuracy (out-of-bag error) and the association accuracy (variable importance) caters for classification as well as feature selection approaches. Furthermore, in case of variable importance, we not only look at changes in the measured importance of each feature but also the rank of each feature in the importance list. We evaluate our method on Bone Mineral Density case/control Dataset and demonstrate how the time can be saved while maintaining the same accuracy.

KEY WORDS

GWAS, Random-Forest

POSTER 7

Unbiased SNP Selection Method for Epistasis Detection in Genome-Wide Association Study (GWAS)

Mr Xiaochuan Wang
Cancer Council Victoria, Melbourne, Australia

BIOGRAPHY:

Harry is Senior Research Officer at Cancer Epidemiology & Intelligence Division in Cancer Council Victoria with background in both computational science and biochemistry

ABSTRACT:

Xiaochuan (Harry) Wang[1], James Chamberlain1, Roger Milne[1]

[1] Cancer Epidemiology & Intelligence Division, Cancer Council Victoria, Melbourne Vic Australia

Currently, genetic variants discovered in GWAS can only explain a fraction of total phenotypic variation, and epistasis may be one of the missing pieces in the puzzle. However, when testing all possible epistasis terms, or even only two-way epistasis, across the entire genome, the computational burden is generally far beyond the practical limitations of current systems. To our knowledge, compared to generalized linear regression based detection models, the majority of currently available computationally optimized epistasis detection algorithms have various drawbacks in the initial SNP candidate selection phase. These drawbacks include being (i) highly hypothesis-dependent and (ii) approximation based, which could result in, at least in part, the genome coverage being too narrow. In order to achieve a hypothesis free epistasis detection using generalized linear regression based models and the best detection accuracy, it is critical to ensure both that the total number of candidate SNPs is manageable and that the subsampling method satisfies three goals: (1) good genome-wide coverage; (2) the method is regionally unbiased and (3) the method is hypothesis-free.

However, most common selection methods cannot meet all these criteria, especially when imputed allele dosage values with different imputation quality scores are involved. Therefore, we have developed a new SNP selection method using the probabilistic pigeonhole principle. Compared to other commonly available methods, it is more flexible in allowing extra parameters, such as imputation quality scores, into the selection process, and it is based on an unbiased statistical model. The selection method is primarily driven by the variants' genome location, combined with other evaluative scores to prioritize the genome-wide coverage. This method is potentially applicable to several scenarios, such as when the data is combined from genotyping arrays with and without genome-wide backbone or from genotyped and imputed SNPs.

KEY WORDS

GWAS, Selection, Unbiased, SNP

POSTER 8

Testing for differential transcript usage in model organisms using superTranscripts annotations

Ms Damayanthi Herath
Optimization and Pattern Recognition Research Group, Melbourne School of Engineering, The University of Melbourne, Parkville, Australia,

BIOGRAPHY:

Damayanthi Herath is a graduate from and an academic staff member of the Department of Computer Engineering, University of Peradeniya, Sri Lanka. She is currently studying for a PhD in the Optimization and Pattern Recognition research group at Melbourne School of Engineering, The University of Melbourne, Victoria, Australia. She seeks to reveal the wonders of ACGT s using 0's and 1's.

ABSTRACT:

Damayanthi Herath[1,2], Alicia Oshlack[3], Nadia Davidson[3]

[1] Optimization and Pattern Recognition Research Group, Melbourne School of Engineering, The University of Melbourne, Parkville, VIC 3052, Australia
[2] Department of Computer Engineering, University of Peradeniya, Peradeniya, Sri Lanka
[3] Murdoch Children's Research Institute, Royal Children's Hospital, Flemington Road, Parkville Victoria 3052 Australia

RNA-Seq is being widely used to study the transcriptome given its advantages over the traditional approaches. One of the applications of RNA-Seq is the analysis of expression of multiple transcripts of a gene across experimental conditions (differential transcript usage). Filtering isoforms with low abundances prior to the analysis has been previously suggested for better control of the false-discovery rate. A key step in RNA-Seq data analysis is aligning the sequence reads to a reference genome. SuperTranscripts is an alternate reference sequence for alignment and annotation that has been proposed for RNA-Seq data analysis in the absence of a complete reference sequence.

This study explored using superTranscripts to test for differential transcript usage in organisms where good reference genomes and annotations are available.
We considered three different superTranscripts annotations including reference based, data driven and a combination. Once the superTranscripts annotations were created, reads mapping to multiple exonic sequences of a gene were counted based on them, using the tool, FeatureCounts. Next, the count-based framework, DEXSeq was used to infer differential transcript usage.
Simulated data of Drosophila and Human were used to evaluate the performances of superTranscripts annotations based approaches and standard methods to analyze differential transcript usage. The proposed methods resulted in better control in false discovery rate than the standard reference based methods. Furthermore, they do not require prior estimation of isoform abundances and resulted in performances comparable to isoform prefiltering methods. The proposed methods were applied to an experimental dataset of Drosophila and were able to detect genes with differential transcript usage that were not detected by standard methods. The analysis of real data demonstrated that they may also be used to visualize the differential transcript usage of genes effectively.

KEY WORDS

SuperTranscripts, Differential Transcript Usage, Model organisms, Visualisation

POSTER 9

Long read de novo assembly for large genomes: experiences from the frontline

Dr Peri Tobias
School of Life and Environmental Sciences, University of Sydney, Eveleigh, Australia

BIOGRAPHY:

Peri Tobias completed her PhD at the University of Sydney in 2017. Her research focus has been the genetic basis of plant resistance to myrtle rust. Her broader research interest is in woody plants and their molecular response to pathogens. Her work crosses molecular biology and bioinformatics.

ABSTRACT:

Peri A Tobias[1,2], Mark Powrie, David Chagne[2], Robert F. Park[1,3]

[1] School of Life and Environmental Sciences, University of Sydney, Eveleigh, NSW 2015, Australia
[2] The New Zealand Institute for Plant & Food Research Ltd., Auckland, New Zealand
[3] Plant Breeding Institute, University of Sydney. Private Bag 4011 Narellan, NSW 2567, Australia

Long read sequence data provides a promising alternative for de novo genome assemblies, particularly when the genome is predicted to include a significant component of highly repetitive sequence. The reality is, however, that the computer resources required for assembling genomes larger than several hundred mega-base pairs (Mbp) can make the process extremely slow. Based on flow cytometry and kmer analysis, the fungal genome we are assembling is estimated at haploid size of around 1 400 Mbp. We obtained raw read outputs from two generations of Pacific Biosciences (PacBio) sequencers; RSII (bax.h5) and Sequel (bam). Fasta and Quiver/Arrow files were extracted, and the assembly process initiated with Canu (v1.6) long read assembly software.

An initial assembly with 30 times read coverage completed within 2.5 months using the University of Sydney high performance compute (HPC) cluster. Contig numbers were high (22 474) and BUSCO analysis indicated that 80% complete and 6.6% fragmented gene models were present. A further eight more SMRT cells (Sequel) were therefore run for greater read depth. It was hoped that a higher coverage of long read sequence data would help to build a more contiguous genome assembly. The additional reads provided a total of 50 to 60 times coverage. A new assembly was initiated for this moderately large genome however this has greatly increased compute resource requirements. The current assembly has been running for more than three months on the HPC cluster. Here, we discuss the problems encountered during the assembly process of such a highly repetitive fungal genome.

POSTER 10

Dysregulation of miRNA Expression in the Peripheral Blood of Patients with Schizophrenia

Ms Behnaz Khavari
School of Biomedical Sciences and Pharmacy and the Centre for Translational Neuroscience and Mental Health Research, University of Newcastle and the Hunter Medical Research Institute, Newcastle, Australia

BIOGRAPHY:

Behnaz is doing her PhD in Medical Biochemistry at the University of Newcastle, Australia. Her research interests include investigating the role of non-coding RNAs in the etiology and diagnosis of neurodevelopmental disorders, specifically Schizophrenia, through the application of RNA sequencing and other high-throughput technologies.

ABSTRACT:

Behnaz Khavari[1], Michael Geaghan[1], Joshua Atkins[1], Dylan Kiltschewskij[1], Vaughan Carr[2,3], Melissa Green[2,3], Murray Cairns[1,3]

[1] School of Biomedical Sciences and Pharmacy and the Centre for Brain and Mental Health Research, University of Newcastle and the Hunter Medical Research Institute, Newcastle, NSW, Australia.
[2] School of Psychiatry, University of New South Wale, Sydney, Australia.
[3] Schizophrenia Research Institute, Sydney, Australia

Schizophrenia (SZ) is a debilitating neuropsychiatric disorder that affects 1% of the population. Patients typically display a broad range of symptoms, including hallucination, loss of affect and severe cognitive impairments. While the diagnosis is currently based on signs and symptoms, by understanding the molecular determinant of SZ, we may be able to identify biomarkers and better direct treatment to the underlying causal factors. Genomic analyses collectively indicate SZ as a complex developmental disorder involving many genes as well as epigenetic factors.

MicroRNAs (miRNAs) are a class of small non-coding RNAs which are sensitive to environmental influences and appear to be associated with the etiology of SZ. The aim of the current study was to explore if there is a miRNA expression signature associated with functionally significant phenotypic subtypes of SZ. We conducted small RNA-seq on peripheral blood mononuclear cells (PBMCs) of male patients with severe cognitive deficits, collected by the Australian Schizophrenia Research Bank (ASRB), and compared it to those with moderate impairments (CS group). Raw sequencing reads were aligned to the reference genome (hg38) and analysed further through a customized pipeline, including the software FastQC, Cutadapt, Bowtie2 and Htseq. Employing the edgeR package of RStudio, we could detect several differentially expressed miRNAs with P-value<0.05. Pathway analysis for the most highly differentiated miRNA, using ToppFun, suggested the molecule is involved in several pathways related to the brain function and development. These pilot data suggest there may be miRNA expression changes associated with the cognitive deficit subtype of SZ to explore as an interesting candidate biomarker worthy of further investigation.

KEY WORDS

Schizophrenia, biomarker, microRNA, peripheral blood mononuclear cells, RNA-Seq

POSTER 12

What are the early changes in brain function driving Alzheimer's disease? Transcriptome-based analyses of familial Alzheimer's disease-like mutations in zebrafish

Dr Michael Lardelli
The University of Adelaide, Adelaide, Australia

BIOGRAPHY:

1987 (September) to 1991 (March), PhD studies (with CNAA) - Imperial Cancer Research Fund Developmental Biology Unit, Oxford, U.K., Regulation and function of the Drosophila pair-rule gene hairy.

1991 (March) to 1994 (July), Postdoctoral studies - The Karolinska Institute, Stockholm, Sweden. Molecular genetic analysis of early mouse neural development.

1994 (July) to 1995 (June), Postdoctoral studies at Uppsala University Biomedical Center, Sweden. Department of Developmental Neuroscience. Genetic studies of vertebrate development using the zebrafish.

1995 (July) to 1997 (May), Assistant Professor at Uppsala University Biomedical Center, Sweden. Department of Developmental Neuroscience. Genetic studies of vertebrate development using the zebrafish.

1997, AMRAD postdoctoral award for biomedical researchers returning to Australia.

1997 (June) to 2003 (June), Lecturer, then 2003 (July) to 2014 (December), Senior Lecturer, then 2015 (January onwards), Associate Professor, at The University of Adelaide, Australia. Developmental genetics studies and then analysis of Alzheimer's disease genetics using the zebrafish.

ABSTRACT:

Nhi Hin[1], Morgan Newman[1], Stephen Pederson[1], **Michael Lardelli[1]**

[1] University of Adelaide, School of Biological Sciences, Centre for Molecular Pathology, North Terrace, Adelaide, SA 5005

Alzheimer's disease (AD) accounts for around 70% of dementia cases. However, despite decades of intensive research, there is still no consensus on the molecular basis of the disease and its cause. While the disease takes many years to develop, detailed molecular analysis of AD brains is only possible using post-mortem tissue. The predominant opinion within AD research is that accumulation of the peptide Amyloid drives disease progression. However, what causes Amyloid to accumulate is uncertain. In any case, much evidence is inconsistent with this hypothesis and it has not led to successful ameliorative treatments. Transcriptomic analysis of transgenic mouse models (that have been constructed mainly on the basis of the Amyloid hypothesis) does not support that these model the disease. We postulate that, in the absence of a firm understanding of the disease mechanism, the most objective and valid approach to modelling the disease is to recreate the human familial AD (fAD) genetic state as closely as possible – in other words, to examine the effects of single, heterozygous fAD-like mutations in endogenous genes. The mouse knock-in models of fAD mutations created over 15 years ago were never analysed using transcriptomics since the technologies did not exist at that time. To reduce genetic and environmental noise in our transcriptome analyses of fAD "knock-in" mutations, we exploit the large sibling numbers and environmental consistency possible with zebrafish culture. We have generated fAD-like mutations in a number of genes in zebrafish and we are analysing their brain transcriptomes as they age. Ultimately, by comparing fAD-like and non-fAD-like mutations in these genes we aim to derive "intragenic" fAD transcriptomic signatures. The fAD signatures of different genes can then be compared to define a common "intergenic" signature that identifies core molecular/cellular changes in AD. Initial results indicate that energy metabolism stress may drive AD pathology.

KEY WORDS

PRESENILIN, SORTILIN-RELATED RECEPTOR, FAMILIAL ALZHEIMER'S DISEASE, ZEBRAFISH, TRANSCRIPTOME

POSTER 13

Sequencing of human genomes with nanopore technology

Dr Rory Bowden
Wellcome Centre For Human Genetics, University of Oxford, Oxford, United Kingdom

BIOGRAPHY:

Rory Bowden has a BSc (Hons) from the University of Adelaide and a PhD from the University of Cambridge. He currently holds a leadership position in a large and diverse genomics core lab at the University of Oxford, where he has special responsibility for scientific collaborations and innovation. He has interests in virus and bacterial genomics, sequencing technologies and single-cell genomics, from practical and analytical viewpoints.

ABSTRACT:

Rory Bowden[1], Robert W Davies[2], Andreas Heger[3], Alistair T Pagnamenta[1], Mariateresa de Cesare[1], Michael Simpson[2], Peter Donnelly[3]

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[2] Genomics plc, Oxford, UK
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Whole-genome sequencing (WGS) is becoming widely used in clinical medicine in diagnostic contexts and to inform treatment choice. While current sequencing technologies have been extremely successful, their reliance on short read lengths necessarily involves some limitations: accurately assaying certain genomic regions and classes of variation can be problematic. Recent advances in throughput and cost have made WGS using the Oxford Nanopore Technologies (ONT) MinION long-read single-molecule sequencer a potential solution to these challenges. Recent publications have demonstrated that ONT data can enable highly contiguous human genome assemblies, without addressing whether ONT data can support variant calling at accuracies sufficient for its use as a standalone clinical sequencing platform. Here we evaluate the potential for routine WGS using ONT by sequencing the well-characterised reference sample NA12878 and the genome of an individual with ataxia-pancytopenia syndrome accompanied by severe immune dysregulation, to 79× and 30× respectively. For NA12878, we evaluated single-nucleotide variant (SNV) calls based on data from multiple base-calling algorithms. We demonstrate that phasing metrics from a novel, reference panel-free, long-read-based method can improve variant-calling performance from otherwise modest levels, resulting in a false discovery rate of 0.079 and false negative rate of 0.085; remaining errors are concentrated near homopolymers and in regions of reduced sequencing coverage. In the clinical sample, we are able to identify and directly phase two non-synonymous de novo variants in SAMD9L, (OMIM #159550) inferring that they both lie on a common paternal haplotype by overlap with parental genotypes at nearby common variants. This work demonstrates that methodological innovation can substantially reduce variant-calling error rates in ONT data, and that with on-going improvements in throughput, base-calling and dedicated long-read-based SNV-calling methodology, ONT offers promise as an option for clinical WGS.

KEY WORDS

WGS, Clinical Sequencing, Genome Assembly

POSTER 14

New Developments in NGS sample Quality Control – from FFPE RNA to cell-free DNA

Dr Dan Belluoccio
Agilent Technologies, Waldbronn, Germany

BIOGRAPHY:

Rainer Nitsche is product manager within the Biomolecular Analysis Division marketing team of Agilent Technologies based in Waldbronn, Germany. He is responsible for the next generation TapeStation instrumentation. His background is chemistry as well as biochemistry as he received his PhD from the faculty of Biochemistry at University of Karlsruhe, Germany.

ABSTRACT:

Rainer Nitsche, Eva Graf, Elisa Viering

Agilent Technologies, Waldbronn, Germany

Over the past years, Next Generation Sequencing (NGS) developed to a powerful tool in almost all genetic research and diagnostic areas. Key for the success of any NGS experiment is the quality control (QC) of source RNA and DNA samples as well as generated libraries. Most library preparations remain lengthy and therefore are expensive processes. Tight QC steps are required to avoid a "garbage in-garbage out" situation. The ideal NGS QC solution is easy-to-use, economical and provides fast and unambiguous results also for very low concentrated samples. One way to ensure that samples are "fit for purpose" is to apply a separation by automated electrophoresis followed by fluorescence detection and automatic data analysis. This talk covers the latest developments in the area of NGS sample QC and gives application examples:

Due to the nature of the material, the degradation level of RNA extracted from formalin fixed paraffin embedded (FFPE) tissue is usually high and a classification with the well-established RNA Integrity number (RIN) frequently not meaningful. Examples will be presented that the DV200 value as parameter for the fragment size distribution is a useful and highly reproducible alternative to this.

An additional example for the importance of QC for NGS workflows is the quality control of cell-free DNA (cfDNA). cfDNA samples are extracted from blood specimens ("liquid biopsies") or other body fluids gain more and more importance in the context of cancer research and prenatal testing. Accurate quantification of cfDNA samples is essential to determine suitable input amounts for cfDNA library preparation prior to sequencing. Dependent on preanalytical sample treatment or extraction method, cfDNA samples may contain larger DNA fragments e.g. genomic DNA contaminations. High molecular weight material can negatively influence library preparation and subsequently result in lower sequencing depth. Different examples for cfDNA samples will be shown and data will be discussed.

KEY WORDS

Next Generation Sequencing (NGS), Quality Control (QC), formalin-fixed paraffin embedded RNA (FFPE RNA), RNA Integrity number (RIN), Degradation value 200 (DV200), cell-free DNA (cfDNA), liquid biopsy, automated electrophoresis

POSTER 15

Direct RNA sequencing of human transcripts using Nanopore sequencing

Dr Lachlan Coin
Institute for Molecular Bioscience, The University of Queensland, Brisbane, Australia

BIOGRAPHY:

Dr. Devika Ganesamoorthy is an early career researcher at the Institute of Molecular Bioscience in University of Queensland. She completed her PhD in 2014 at University of Melbourne. Her research is primarily focused on development and assessment of high throughput methods to analyse genomic variations. She has extensive experience with Nanopore long read high throughput sequencing technology and has explored the method for various applications. She also has extreme interest in the analysis of cell-free DNA for biomarker discovery in various applications.

ABSTRACT:

Devika Ganesamoorthy[1], Hyun Jae Lee[2], Antje Blumenthal[3], Luregn Schlapbach[4] and **Lachlan Coin[1]**

[1] Institute for Molecular Bioscience, The University of Queensland, Brisbane, Australia
[2] QIMR Berghofer Medical Research Institute, Brisbane, Australia
[3] The University of Queensland Diamantina Institute, The University of Queensland, Brisbane, Australia
[4] Lady Cilento Chidrens Hospital, Mater Research, Brisbane, Australia

Direct RNA sequencing analysis is currently feasible using Nanopore sequencing and it has opened new frontiers in transcriptomic analysis. Long read sequencing combined with native RNA sample analysis provides opportunity to analyse known full length transcripts, isoforms and discover novel transcripts, isoforms and fusion events. Furthermore, native RNA analysis removes the need for conventional cDNA preparation, hence eliminates the PCR bias frequently observed in isoform analysis. We utilized blood samples collected in PAXgene tubes to assess the utility of direct RNA sequencing for human transcriptomic analysis.

Total RNA without polyA enrichment was used as input for Nanopore sequencing analysis. Direct RNA sequencing and direct cDNA sequencing were performed using Oxford Nanopore MinION sequencing. Short read Illumina RNA sequencing data were also obtained for these samples.

We generated more than 300,000 reads per sample for direct RNA sequencing. Despite low coverage, greater than 60% of the reads aligned to human genome and had 80% sequence read identity. The correlation in read count at gene level between Nanopore direct RNA and direct cDNA was 0.6 and correlation between Nanopore direct RNA and Illumina cDNA was 0.6. We identified full length transcripts, isoforms and fusion gene events in direct RNA sequencing data, which were confirmed either by Nanopore direct cDNA or Illumina RNA sequencing data or by both. We observed an enrichment in 3` of the transcripts in direct RNA sequencing data, elucidating possible underlying biological mechanisms.

We have successfully generated direct RNA sequencing data on RNA extracted from PAXgene tubes using Nanopore sequencing. Our results demonstrates it is feasible to generate direct RNA sequencing data comparable to cDNA sequencing data. We believe utilizing direct RNA sequencing for transcriptomic analysis has the potential to enhance our understanding of human transcriptomics and improve discovery of transcriptomic biomarkers.

KEY WORDS

Direct RNA sequencing, Nanopore sequencing, transcript, isoforms

POSTER 16

Hyb & Seq™ Technology: A no amplification, no library, single molecule sequencer designed for future clinical application

Dr Michael Rhodes
Nanostring Technologies, Seattle, United States

BIOGRAPHY:

Graduated from York University with a degree in Genetics, did a Ph.D. in Bioinorganic Chemistry at University of London. After a post doc in Chicago working on genetics of metal transport in P. aeruginosa, returned to UK to work at United Kingdom Human Genome Mapping Project Resource Centre finishing as Operation Manager in charge of four teams: - Mouse resequencing, linkage Hotel, Academic Services and Custom services. Joined Applied Biosystems in 1999, worked on Genotyping, qPCR and finally Next Generation Sequencing. Joined Nanostring in 2012 after seeing the potential of the nCounter technology to take the discoveries from NGS and apply them to translational research. At Nanostring he has worked on many new applications, including 3D biology and advanced analysis for nSolver and is now helping develop Hyb & Seq™.

ABSTRACT:

Michael Rhodes[1], Hyb & Seq Team[1]

[1] Nanostring Technologies, Seattle WA USA

Hyb & Seq™ technology is an innovative library-free, amplification-free, single-molecule sequencing technique that uses cyclic nucleic acid hybridization of fluorescent molecular barcodes onto native targets. Hybridization-based sequencing enables the simplest sample-to-answer workflow; both DNA and RNA are directly sequenced with almost no manipulation of the input material. The advantages of single molecule sequencing include simple error correction and digital counting to elucidate DNA copy numbers and RNA levels.

In this presentation we will describe a 24-hour sample to answer workflow that includes;

- Simple direct sample preparation including lysis and sample purification directly from formalin-fixed paraffin embedded (FFPE) tissue.
- Total time from sample to loading on sequencer is under 60 minutes, with total hands-on time of less than 15 minutes, most of this process is now automated
- Simultaneous capture of DNA gene targets and mRNA from with no PCR amplification and no cDNA conversion
- Panel based Targeted sequencing carried out on a microfluidic cartridge.
- Results from a representative set of clinical oncology variants including clinically relevant SNVs and Indels (2-18 bases) which was tested on FFPE clinically prepared samples. Sequencing accuracy reached 99.99% (QV40) when a base from a single molecule was read > 5 times. Targeted variants as low as 1% were successfully detected
- Results from an infectious agent panel including Yeast, gram positive bacteria, gram negative bacteria and virus, simultaneously tested with ability to detect as low as 10 genome copies
- A unique data analysis scheme using a Hyb & Seq assembly algorithm (ShortStack™) for variant calling will be described

Hyb & Seq technologies simplicity, flexibility, and accuracy offers an ideal sample-to-answer solution for the translational sequencing research lab.

Hyb & Seq technology is intended for Research Use Only. It is not for use in any diagnostic procedures.

KEY WORDS

Next generation Sequencing, New Technologies

POSTER 17

Controlling the Outcome of CRISPR-Cas9 Cleavage via Prediction of Microhomology-mediated DNA Repair

Dr. Eva Chan
CSIRO, Redfern, Australia

BIOGRAPHY:

Dr Eva Chan completed her PhD in Bioinformatics at the University of New South Wales, where she studied the Genetic Influence of Gene Expression. Following two Bioinformatics postdoctoral positions, at CSIRO (QLD, Australia) and University of California, Davis (CA, USA), Dr Chan took up a position as the Statistical Genetics Lead at the Vegetable Seeds Division of Monsanto Company, in California, in 2009.

Returning to Sydney in 2013, Dr Chan joined the Garvan Institute of Medical Research, undertaking research in human diversity with particular interest in large-scale structural variations.

ABSTRACT:

Marc Horlacher[1], Laurence Wilson[1], Denis Bauer[1]

[1] CSIRO Health & Biosecurity

The CRISPR-Cas9 System is extensively used in a variety of different fields of research as it allows for a precise generation of DNA double-strand breaks (DSB) at specific genomic loci. However, factors determining the repair pathway of choice are poorly understood, leading to largely unpredictable mutational outcomes and hindering the advancements of the technology into fields with high precision requirements, such as clinical applications.

One repair pathway, Microhomology-mediated End-joining (MMEJ), utilizes hybridization of short homologous sequences up-and downstream of the DSB, leading to repair of the DSB but loss of the intermediate sequence. Knowledge of the homologous sequences utilized allows for an unambiguous characterization of the resulting deletion. Thus, leverage of the MMEJ repair pathway has the potential to precisely control the mutational outcomes of CRISPR-Cas9 experiments.

Previous methods for prediction of MMEJ-associated repair suffer from their reliance on mostly arbitrary cutoffs. Using next-generation sequencing data and a machine learning approach, we predicted the likelihood of DSBs at specific genomic loci being repaired through MMEJ. By comparing mutational outcomes of target-sites treated with CRISPR-Cas9, we identified essential features for MMEJ-associated repair and build a model for classification of target-sites into MMEJ-active and inactive as well as ranking of potential homologies according to their likelihood of utilization. To our knowledge, this is the first solely data-driven method for prediction of MMEJ-associated repair.

We believe that our work will aid researchers in the design of optimal CRISPR-Cas9 target sites by allowing for a more refined selection of suitable genomic loci through prediction of MMEJ-associated mutational outcomes.

KEY WORDS

CRISPR, Machine Learning, DNA Repair, MME

POSTER 2

High-Depth Whole Genome Sequencing of 696 Vascular Plants from one Botanical Garden

Dr Huan Liu
BGI Shenzhen, Shenzhen, China

BIOGRAPHY:

Dr. Huan Liu is a research scientist focused on plant genomics. His work is committed to molecular biology, genetic engineering, genomics and other biogenetic studies. He is primarily responsible for the Ten Thousand Species of Plant Genomics Project (10KP), the African Orphan Crop Genome Project, herb-omics research, and new species development. He has published 15 papers and applied for 22 patents among which 7 have been granted. Dr. Liu also co-authored a book and has won a number of awards and honours including High-level Talents in Biology Industry of Yantian District, Shenzhen City and Top Research Award of the Chinese Elite Scientific Journals (F5000).

ABSTRACT:

Huan Liu[1,2], Ting Yang[1,2], Weixue Mu[1,2], Xin Liu[1,2]

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Genome sequencing has been widely used in plant research to construct reference genomes and provide evolutionary insights. However, only a limited number of the plant species has its whole genome sequenced, and the limited taxon information of these species further restrain the use of these data. Here, we comprehensively sampled and sequenced vascular plant species of Ruili Botanical Garden, located at southwest China. In total, we sequenced 760 samples out of the total 1,093 samples which we also remained images and specimens. These 760 samples represented 696 vascular plant species from 135 families belonging to 48 orders. We generated 54 Tb sequencing data in total, which resulted in an average sequencing depth of 60× for these species, as estimated by the genome size. We then validated the quality and possible applications of the data through establishing the chloroplast genomes of these species and constructing a phylogenetic tree of vascular species. In this study, we established a large dataset of vascular plants, with both the high-depth whole genome sequencing data and the voucher specimen, making it valuable dataset for plant genome researches and applications.

KEY WORDS

Ruili Botanical Garden, Vascular Plant Species, Genome Sequencing

POSTER 3

Detection of dicistronic tRNA:mRNA transcripts in grapevine (Vitis vinifera)

Na Sai
(presenting on behalf of Rakesh David)
School of Agriculture Food and Wine, University of Adelaide, Adelaide, Australia

BIOGRAPHY:

Rakesh David is a Postdoctoral Research Fellow at the ARC Centre of Excellence in Plant Energy Biology, The University of Adelaide.

Areas of research include epigenetics, RNA biology, computational biology
Detection of dicistronic tRNA:mRNA transcripts in grapevine (Vitis vinifera)

ABSTRACT:

Pastor J Fabres[1], Na Sai[1,2], Fei Zheng[1], Stephen M Pederson[3], Penny J Tricker[1], Matthew Gilliham[1,2], Steve Tyerman[1,2], Cassandra Collins[1], Carlos Rodriguez Lopez[4], Rakesh David[1,2]

[1] School of Agriculture Food and Wine, University of Adelaide, South Australia, Australia
[2] ARC CoE in Plant Energy Biology, University of Adelaide, South Australia, Australia
[3] Bioinformatics Hub, University of Adelaide, South Australia, Australia
[4] Environmental Epigenetics and Genetics Group, College of Agriculture, Food and Environment, University of Kentucky, USA

Nuclear-encoded genomic features such as tRNAs and mRNAs are generally expressed as single transcriptional units in plants. However, a recent study demonstrated tRNA and mRNA genes are frequently produced as dicistronic transcripts in the model plant species Arabidopsis. A signal encoded within these tRNA:mRNA genes also triggers systemic transport of these transcripts to distant plant cells. It is not known, however, the extent to which these unusual transcripts are prevalent in other plant species and whether environmental or developmental factors contributes to their regulation.

Here, we designed a customized bioinformatic pipeline to detect and statistically verify dicistronic transcripts between annotated tRNAs and protein-coding mRNAs. Deep sequencing of RNA from leaf tissue of grapevine plants (Vitis vinifera, cv Shiraz) growing in six regions of the Barossa Valley (South Australia) revealed 14% of all detected tRNAs were co-transcribed with their neighbouring mRNA gene (adj. $P < 0.05$). Majority of all dicistronic transcripts were detected in more than one grape-growing regions of the Barossa Valley with only a few that were region-specific. Furthermore, the average inter-gene distance between the tRNA and the adjacent gene was 300 base pairs and we found no directional bias to the transcript.

To understand the developmental regulation of these transcripts, publicly available RNA-seq data from multiple grapevine tissues was also analysed. tRNA:mRNA transcripts were detected in grapevine leaves, roots, flowers and berries with over 50% of the transcripts showing distinct profiles suggesting possible tissue-specific regulation. Future studies will focus on detection of tRNA:mRNA transcripts in range of plant species which will provide clues to the evolutionary conservation of these transcripts and their prevalence in the plant kingdom.

KEY WORDS

Dicistronic transcript, transfer RNA, Vitis vinifera

POSTER 5

Comprehensive HIV surveillance and clinical monitoring at scale: Optimised, low-cost, whole-virus-genome sequencing is ALL you need.

Dr Rory Bowden
Wellcome Centre For Human Genetics, University of Oxford, Oxford, United Kingdom

BIOGRAPHY:

Rory Bowden has a BSc (Hons) from the University of Adelaide and a PhD from the University of Cambridge. He currently holds a leadership position in a large and diverse genomics core lab at the University of Oxford, where he has special responsibility for scientific collaborations and innovation. He has interests in virus and bacterial genomics, sequencing technologies and single-cell genomics, from practical and analytical viewpoints.

ABSTRACT:

David Bonsall[1], Tanya Golubchik[2], Mariateresa de Cesare[3], Christophe Fraser[4], **Rory Bowden[3]**

[1] Big Data Institute and Wellcome Centre for Human Genetics, University of Oxford, UK
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[4] Big Data Institute, University of Oxford, UK

Virus sequencing provides an essential tool in combatting HIV infection at the individual and population level by monitoring the spread of drug resistance, directing optimal antiretroviral regimes, and inferring transmissions and transmission dynamics. Next-generation, whole-genome sequencing provides clear advantages in throughput and resolution, but efforts to sequence HIV genomes at scale face three major technical challenges: (i) minimising assay cost and protocol complexity, (ii) maximising sensitivity, and (iii) recovering accurate and unbiased sequences of both genome consensus and within-host viral diversity.

We have implemented a novel, high-throughput, virus-enrichment sequencing method and computational pipeline tailored to HIV (veSEQ-HIV), which addresses these challenges and can be used directly on blood left over from routine CD4 testing. In a large proof-of-concept trial of 1,620 plasma samples, of which 10% had had viral loads (VLs) measured using the standard clinical qPCR test, we recover complete HIV genomes from a variety of subtypes and 93% of samples with VL > 1,000 copies / ml. The close relationship between yield of HIV reads and VL demonstrates that our method can replace standalone clinical VL testing in many settings, and implies that within-sample variant frequencies estimated with veSEQ-HIV closely represent true variant frequencies previously estimated by sequencing sub-genomic amplicons. Our method and computational pipeline optimises sequencing insert length for the detection of multiple successive minority variants in individual reads from across the genome, a source of information that we exploit for inference of transmissions and prediction of drug resistance. We use updates to the phyloscanner software (phyloscanner clean) to counter a widespread artefact of Illumina sequencing, identifying and excluding illicit reads from other simultaneously processed samples with 95% sensitivity and 99% specificity. veSEQ-HIV compares favourably in cost with a single conventional HIV load, genotyping or drug resistance assay, making it well-suited to large-scale public health efforts.

A preprint containing complete information about the study, authors and affiliations is available at https://www.biorxiv.org/content/early/2018/08/23/397083.

KEY WORDS

HIV, Viruses, WGS, RNA Sequencing, Public Health, Epidemiology, Infectious Diseases

POSTER 6

Evaluation of internal controls on V1-V3 & V3-V4 primer sequences across various AGRF nodes on different MiSeq machines.

Mr Naga Kasinadhuni
Australian Genome Research Facility, St Lucia, Australia

BIOGRAPHY:

Naga Kasinadhuni works as bioinformatics Officer at Australian Genome Reserach facility. As a primary analyst, works on metagenomics, genotype by sequencing, SNP & microsatellite discovery projects, and as secondary analyst works on denovo assembly, RNA seq analysis projects. Has a B.Tech degree in Industrial biotechnology from Anna University, India. Also has a masters degree in Biotechnology & Bioinformatics from La Trobe University, Melbourne.

ABSTRACT:

Naga Kasinadhuni[1], Christopher Noune[1], Justin Crockett[1], Kenneth Chan[1]

[1] Australian Genome Research Facility, Australia

Understanding microbial diversity is important in microbial studies. Traditional cultivation-dependent methods have been a main bottleneck for these studies. The advancement of high-throughput sequencing platforms has revolutionized our ability to investigate the microbiota composition of complex environments. One of the most popular approach in studying microbial community composition is the 16S rRNA gene targeted sequencing. Numerous factors, including DNA extraction method, primer sequences and sequencing platform employed, can affect the accuracy of the results. Therefore, many studies strongly advocate the use of internal controls. In Australian genome research facility (AGRF), we provide microbial diversity profiling service for 16S rRNA gene using V1-V3 (27F:519R) and V3-V4 (341F:806R) primer sequences.

While sequencing client samples on different MiSeq machines across various nodes including Brisbane, Melbourne and Adelaide, we use the HM-782D from bei Resources as an internal control. The aim of this study is to evaluate the consistency of the microbial diversity profiling by comparing the internal controls across various nodes on different MiSeq machines over a span of one year. The results show remarkable consistency of the internal controls across various nodes on all MiSeq sequencing machines.

KEY WORDS

Metagenomics, Bioinformatics, controls

POSTER 7

Deep Sequencing of Microbial Communities in Cystic Fibrosis Airways

Ms. Tania Duarte
Institute for Molecular Bioscience, The University of Queensland

BIOGRAPHY:

Tania graduated with a Bachelor's and Master's degree in Biochemistry at the University of Coimbra, Portugal. She is now a second year PhD candidate at the A/Prof Lachlan Coin's group -Institute for Molecular Bioscience, The University of Queensland, Australia - where she previously worked as research assistant. Her research interests include high throughput sequencing, specially Oxford Nanopore technologies sequencing platforms, bioinformatics, metagenomics and microbiology of CF patients airways.

She has many other interests, such as photography and dance.

ABSTRACT:

Tânia Duarte[1], Lachlan Coin[1], Erin Price[2], Derek Sarovich[2], Sanjaya Kc[1], Son Nguyen[1], Thuy-Khanh Nguyen[3] and Scott Bell[3,4,5]

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[2] Faculty of Science, Health, Education and Engineering, University of the Sunshine Coast, Sippy Downs, QLD 4556 Australia;
[3] QIMR Berghofer Medical Research Institute, Herston, QLD 4006 Australia;
[4] Metro North Hospital and Health Service, Royal Brisbane and Women's Hospital, Herston, QLD 4006 Australia;
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Cystic fibrosis (CF) is the most common life-shortening genetic disorder in Australians, with a prevalence of 1 in 2,890 births. CF is caused by mutations in the CF transmembrane conductance regulator gene that result in a dysfunctional protein. Dysregulation of the exocrine system occurs as a consequence, leading to several clinical outcomes, such as chronic lung disease, pancreatic insufficiency and intestinal obstruction.

Lung disease causes the highest rate of mortality in CF patients; however, it remains unclear how microbes can deeply infect the airways and why the immune response is ineffective in eradicating them. Traditional culture methods for pathogen identification are laborious and relatively insensitive, especially for fastidious or slow-growing organisms, and for detecting strain mixtures. Therefore, it is essential to perform deep ("metagenomic") sequencing of the microbial communities to understand their composition and to identify potential links between persistence and antimicrobial resistance (AMR).

This study describes preliminary metagenomic sequencing of total DNA derived from sputum (non-invasive method) of two CF patients using the Oxford Nanopore Technologies (MinION) and Illumina platforms. Despite only a small fraction (~1%) of reads corresponding to non-human DNA, we detected Pseudomonas aeruginosa as the most abundant pathogen, consistent with culture. In addition, an AMR gene (aph(3')-IIb) was identified in 1 patient despite overwhelming human contamination. The detected bacterial infection and AMR gene were validated via culture-based metagenomic sequencing. We are now optimising the DNA extraction protocol to incorporate a human DNA depletion step to enhance detection sensitivity.

Our results provide exciting preliminary data showing that metagenomic sequencing is a powerful method for characterising microbial populations in the CF airway microbiome. Future applications will use metagenomics to examine microbial changes over time and in response to treatment, with the aim of better understanding microbial populations, persistence and evolution, and ultimately, informing patient treatment.

KEY WORDS

Cystic fibrosis, microbiology, antimicrobial resistance, high throughput metagenomic sequencing, bioinformatics

POSTER 8

Identification and Characterisation of Ruxolitinib Resistant Mutations in *JAK2*-rearranged B-cell Acute Lymphoblastic Leukaemia

Ms Charlotte Downes
South Australian Health and Medical Research Institute (SAHMRI), Adelaide, Australia

BIOGRAPHY:

PhD student at the University of Adelaide in the acute lymphoblastic leukaemia research group at SAHMRI.

ABSTRACT:

Charlotte EJ Downes[1,2], Barbara McClure[1,3], Benjamin Mayne[1], John B Bruning[2], Susan L Heatley[1,3,7], Chung H Kok[1,3], Timothy P Hughes[1,3,5], David T Yeung[1,3,5], Deborah L White[1,2,3,4,6,7]

1 Cancer Theme, South Australian Health and Medical Research Institute (SAHMRI), Adelaide, SA, Australia.
2 School of Biological Sciences, University of Adelaide, SA, Australia.
3 School of Medicine, University of Adelaide, Adelaide, SA, Australia.
4 School of Pediatrics, University of Adelaide, Adelaide, SA, Australia.
5 Hematology Department, SA Pathology, Adelaide, SA, Australia.
6 Australian Genomics Health Alliance (AGHA).
7 Australian and New Zealand Children's Oncology Group (ANZCHOG).

Janus kinase 2 (JAK2) rearrangements (JAK2r) occur in approximately 5% of paediatric B-cell acute lymphoblastic leukemia (B-ALL) cases and are associated with poor prognosis. A clinical trial is currently assessing the Jak1/2 inhibitor, ruxolitinib (rux) in high-risk B-ALL cases harbouring JAK2 pathway alterations. Elucidating mechanisms of rux resistance in JAK2r B-ALL will enable the development of therapeutic strategies to overcome or avert resistance. JAK2r B-ALL was modelled in the pro-B cell line, Ba/F3, by expressing the high-risk B-ALL fusion, ATF7IP-JAK2. Rux resistance was generated following dose escalation to a clinically relevant dose of 1 μM in three independent experiments. mRNA sequencing and Sanger sequencing of RT-PCR amplified JAK2 fusion specific transcript revealed each resistant line had acquired a different mutation within the JAK2 kinase domain, suggesting that mutation-based resistance was stochastic. In addition to the identification of two known rux resistant mutations, JAK2 p.Y931C and p.L983F, a novel p.G993A mutation was also detected. Computational modelling of acquired JAK2 mutations and their influence on rux binding was performed using ICM-Pro (Molsoft L.C.C.). The mutations localised to the ATP/rux binding site of the kinase domain and were predicted to reduce rux binding affinity by disruption of critical bonds within the ATP-binding site. mRNA sequencing and gene expression analysis of total RNA from naïve, DMSO-treated (vehicle control), and rux resistant ATF7IP-JAK2 Ba/F3 revealed that overall gene expression differed greatly between the vehicle control and rux resistant cell lines. JAK2 p.Y931C was also observed to have largely different changes in gene expression when compared to the JAK2 p.G993A and p.L983F mutant cell lines. Understanding mechanisms of rux resistance, as modelled here, has the potential to inform future drug design and identify novel therapeutic strategies for this high-risk patient cohort.

KEY WORDS

Acute lymphoblastic leukaemia, Janus kinase 2, resistance, ruxolitinib

POSTER 9

Malignant Pleural Mesothelioma profiling through Single-Cell RNA-seq

Dr Hyun Jae Lee
QIMR Berghofer Medical Research Institute, Herston, Australia

BIOGRAPHY:

Hyun Jae Lee is a research officer in QIMR Berghofer Medical Research Institute studying both cancer and malaria by employing single-cell RNA-seq approach. His PhD involved understanding disease pathogenesis through RNA-seq, including employment of dual RNA-seq approach to study host-pathogen interactions in severe malaria.

ABSTRACT:

Hyun Jae Lee[1]*, Pamela Mukhopadhyay[1]*, Stephen Kazakoff[1], Venkat Addala[1], Michelle Wykes[1], Paul Collins[1], Scott Wood[1], Jessica Engel[1], Siok Tey[1], Ashraful Haque[1], John V Pearson[1], Bruce Robinson[2], Jenette Creaney[2], Ann-Marie Patch[1], Nicola Waddell[1]

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* These authors contributed equally to this work

Malignant pleural mesothelioma (MPM) is a rare but aggressive cancer that originates from the pleural lining around the lungs and is associated with exposure to asbestos. Mesothelioma is known to be a heterogenous cancer. Here, we employed single-cell RNA sequencing (scRNA-seq) to perform high resolution transcriptome analysis of mesothelioma effusions from 3 patients. A high-throughput, low-depth Chromium scRNA-seq approach allowed analysis of 7622 cells. We identified a total of 288 cancer cells that displayed common signatures including MSLN. Most of the non-cancer cells were identified as different populations of immune cells. The single cell data was used to infer copy number variation (CNV) of the cancer cells and revealed intratumoural heterogeneity.

The inferred CNV from single-cell transcriptomic data also showed good correlations with CNV detected from bulk whole-genome sequencing of the same patients. These results illustrate that scRNA-seq is a powerful approach to characterise the heterogeneity of MPM.

KEY WORDS

Cancer, Single-cell RNA-seq, WGS

POSTER 11

Genetic regulation of transcription and methylation in human endometrium and identification of gene targets for reproductive diseases

Sally Mortlock
IMB, The University of Queensland, St Lucia, Australia

BIOGRAPHY:

I completed my PhD in Veterinary Science in 2017 at the University of Sydney. My PhD studies were focused on canine cancer genomics and analysis of omic datasets to identify genomic regions associated with lymphoma in dogs. I am currently a postdoctoral researcher in the Genomics of Reproductive Disorders research group lead by Professor Grant Montgomery at the IMB at the University of Queensland. Our group studies genetic risk factors for reproductive disorders and investigates the functional mechanisms contributing to disease. We are currently focused on identifying and understanding genetic factors contributing to endometriosis risk, understanding genetic and epigenetic regulation of gene expression in the endometrium and conducting functional studies in endometrium to identify target genes. My work within the group centers around statistical and computational analysis of omic datasets and bioinformatics.

ABSTRACT:

Sally Mortlock[1], Restuadi Restuadi[1], Rupert Levien[1], Jane E. Girling[2,4], Sarah J. Holdsworth-Carson[2], Martin Healey[2], Wan Tinn Teh[2], Zhihong Zhu[1], Ting Qi[1], Yang Wu[1], Samuel W. Lukowski[1], Brett D. McKinnon[1,3], Jian Yang[1], Joseph E. Powell[1], Allan F. McRae[1], Peter A.W. Rogers[2], Jenny N. Fung[1], and Grant W. Montgomery[1]

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Multi-omic data integration can be used to unravel complex biological systems and better understand mechanisms regulating gene expression and function in tissues and disease states. Human endometrium is a highly specialised tissue essential for establishment and maintenance of pregnancy and considered a source of cells initiating lesions in endometriosis. Endometriosis occurs when tissue, similar to the endometrium, forms lesions outside the uterus, affecting 10% of reproductive aged women. Genome-wide association analyses have identified 27 endometriosis risk loci. Specific gene targets and genetic mechanism's behind the disease remain to be identified.

Endometrium undergoes dynamic molecular changes across the menstrual cycle in response to steroid hormones and epigenetic modifications. To better understand mechanisms underlying gene regulation in endometrium we analysed both gene expression and methylation and mapped expression quantitative trait loci (eQTLs) and methylation quantitative trait loci (mQTLs) in endometrial tissue from European women. Integrating individual genotype data, we observed a total of 469 sentinel cis-eQTLs ($P < 3.3 \times 10–9$), 104 sentinel trans-eQTLs ($P < 5.4 \times 10–13$), and a total of 4,546 sentinel cis-mQTLs ($P < 1.13$ x 10-10) and 434 sentinel trans-mQTLs ($P <$ 2.29 x 10-12) in endometrium. More than 60% the cis-eQTLs and cis-mQTLs identified in endometrium were also observed in blood.

Approximately 7% of CpG sites showed changes across the menstrual cycle in endometrium and 25% of the genes closest to these sites were also differentially expressed. SMR analysis highlighted potential causative variants associated with expression and/or methylation in endometrium for known endometriosis risk loci VEZT, GREB1 and LINC00339. Endometrial eQTL and mQTL signals also overlapped genomic regions associated with other reproductive traits including endometrial cancer, polycystic ovary syndrome, ovarian cancer, and age of menarche and menopause. By integrating omic datasets we identify strong genetic effects on transcription and methylation in endometrium extending our understanding of tissue specific genomic regulation and providing a platform for better understanding of genetic effects on endometrial-related traits and pathologies.

KEY WORDS

Gene expression, Expression quantitative trait loci (eQTL), DNA methylation, DNA methylation quantitative trait loci (mQTL), Endometrium, Blood, Menstrual cycle, Endometriosis

POSTER 12

Identification of Synergistic Patterns of Genetic Variation that Contribute to Disease Aetiology

Dr Aaron Casey
South Australian Health and Medical Research Institute, Adelaide, Australia

BIOGRAPHY:

Dr Casey completed his PhD in Genetics at the University of Adelaide in 2015 and is currently an EMBL Australia post-doctoral researcher in the Makinen group at the South Australian Health and Medical Research Institute. He currently works on high-throughput pathway analysis using a combination of public resources and in-house software

ABSTRACT:

Aaron E. Casey[1,2] and Ville-Petteri Makinen[1,2]

[1] South Australian Health and Medical Research Institute
[2] School of Biological Sciences, The University of Adelaide, Adelaide, South Australia

Common complex diseases arise from the synergistic combination of genetic risk, environmental factors and aging. Genetic studies indicate a complex aetiology with a large number of variants across the genome influencing disease risk, however, the effect of a single variant tends to be modest. Standard genetic analysis, including GWAS, are not designed to detect the synergistic patterns of genetic variation as they typically focus on single variant contribution to disease aetiology. Our mission is to identify the synergistic patterns of genetic variation that contribute to disease susceptibility. We have developed methodology to integrate pathways and gene-gene interaction networks with genetic signals from GWAS, thus enabling us to ascertain when multiple genes in a pathway/network are perturbed. For our analysis we have collated 693 canonical pathways, 491 drug signature gene sets, 129 disease gene sets (each containing 20-1000 genes) and 1,241 GWAS from 143 sources.

As a positive control we analysed an LDL GWAS where the top gene sets were: 1) canonical pathways – lipoprotein pathways, 2) drug signatures – lipid lowering drugs, and 3) disease pathway – stroke and cardiovascular disease. Interestingly when we analysed a glycine GWAS, we found that the top gene sets were 1) canonical pathways – events in specific receptor tyrosine kinase signalling (receptor tyrosine kinases known to have a role in breast cancer) 2) drug signatures – anticancer drugs 3) disease pathway – polycystic ovary syndrome and breast cancer indicating there is a possible correlation between circulating glycine and breast cancer. We have also recently published a genetic association that links the autophagic and endo-lysosomal system with Alzheimer's disease and we are currently exploring the potential mechanisms to explain the finding. This highlights our ability to use our method in a high-throughput and meaningful manner to create testable hypothesis for experimental studies and drug discovery.

POSTER 13

Detectability of Clinically Important DNA Sequence Variants in Cancer

Mr Peter Tsai
The University of Auckland, Grafton, New Zealand

BIOGRAPHY:

My current research is aimed to use bioinformatics to maximise the amount of biologically and clinically useful information that can be extracted from large cancer genomics datasets. Focusing on the appropriate processing of cancer genomic data and the use of databases containing biological, genomic and clinical information as an informative context for data interpretation.

ABSTRACT:

Peter Tsai[1], Annette Lasham[1], Brett Ammundsen[2], Chris Duran[2], Steven Stones-Havas[2], Cristin Print[1]

[1] Department of Molecular Medicine and Pathology, Faculty of Medicinal and Health Sciences, The University of Auckland, New Zealand
[2] Biomatters Limited, Auckland, New Zealand

The bioinformatic process of somatic DNA sequence variant detection in cancer can be broadly broken down into two steps; first alignment of individual short sequence reads against a reference genome, then variant identification. These steps result in a list of variants that are then filtered and annotated to remove potential 'false positives' and to prioritise the remaining variants for clinical and biological interpretation. However, due to the presence of repeat regions and highly homologous genes, a significant proportion of reads will not be mapped back to the reference human genome uniquely (multiple-mapped reads), which has a direct effect on the accuracy of identifying variants. Here, using in silico approach, we evaluated detectability of coding SNV (single nucleotide variants) in the COSMIC (Catalogue Of Somatic Mutations In Cancer) and CLINVAR variant databases with two well-known somatic workflows (GATK MuTect2 and VarScan2 somatic calling) to identify clinically and biologically important variants that cannot be readily detected using current methods. We identified 48,223 (out of 3,432,727) coding SNV in COSMIC v81 and 1,779 (out of 269,269) coding SNVs in CLINVAR that are not detectable (false negative) using simulated short-read data. Most of these false negative variants are found within known complex repetitive genomic regions in the human reference genome. Since these undetectable DNA variants include many that are clinically important, this illustrates the importance of understanding detection limits when using short DNA sequence reads as the source of evidence in a diagnostic or translational research environment. Clinically important "undetectable" variants need to be highlighted during downstream variant filtering stage for both DNA and RNA variant calling to indicate potential "missed" diagnosis.

KEY WORDS

Somatic, Cancer, Mappbility, False Negative

POSTER 14

VDJPuzzle: A bioinformatics pipeline for simultaneous single-cell characterization of surface phenotype and gene expression profile of T-cell and B-cell from scRNAseq

Ms Money Gupta
Kirby Institute, Viral Immunology Systems Program (VISP), University of New South Wales, Sydney, Australia

BIOGRAPHY:

VDJPuzzle: A bioinformatics pipeline for simultaneous single-cell characterization of surface phenotype and gene expression profile of T-cell and B-cell from scRNAseq

ABSTRACT:

Simone Rizzetto[1,2*], Jerome Samir[1,2*], **Money Gupta[1,2*]**, Willem Van der Byl[1,2], Curtis Cai1,2, Auda Eltahla[1], Rowena Bull[1], Mandeep Singh[3], Joanne Reed[3], Chris Goodnow[3], Fabio Luciani[1,2]

* Equally contributed
1 Kirby Institute, Viral Immunology Systems Program (VISP), University of New South Wales, Sydney, Australia
2 Systems Medicine in Infectious Diseases, University of New South Wales, Sydney, Australia
3 Department of Immunology, Garvan Institute of Medical Research, Darlinghurst, Australia

Lymphocyte recognition of foreign antigens is mediated by the T and B cell receptors (TCR and BCR). Upon antigen recognition, naïve T and B cells proliferate and differentiate, generating a progeny with different phenotypes and transcriptomes, but with same antigen specificity. Measuring phenotype and transcriptional profile, as well as the TCR and BCR sequences, is critical to understanding the immune response and the establishment of protective immunity in acute and chronic infections. The rise of multi-omics approaches such as single cell transcriptomics and proteomics inevitably requires extensive computational workflows specifically designed for immune cells to analyse and integrate large and multiple datasets.

We have developed a computational pipeline (VDJpuzzle) to reconstruct full-length TCR and BCR sequences from single-cell RNAseq (scRNA-seq) data which also allow the identification of receptor isotype and quantified somatic hypermutations along with membrane versus secretory BCR prediction. Immune receptor information is linked to the transcriptional and surface protein profiles obtained from scRNA-seq and flowcytometry, respectively.

This pipeline has been applied to Ag-specific T and B cells derived from human peripheral blood mononuclear cells (PBMC) collected from patients infected with hepatitis C virus, revealing a surprising level of heterogeneity within the same lymphocyte lineages. Our computational analysis contributes to the understanding of the evolution of lymphocytes during an immune response both at the population and at the clonal level, providing insight into the dynamics of immune responses

POSTER 15

Variable Depth Forest: A More Random Random-Forest for heterogeneous disease genetics

Dr Natalie Twine
CSIRO, Australia

BIOGRAPHY:

Arash Bayat is a PhD student at School of Computer Science and Engineering of University of New South Wales (UNSW). He joins Transformational Bioinformatic Team of CSIRO as Postdoctoral fellowship. His research interest are algorithms and bioinformatics.

ABSTRACT:

Arash Bayat[1], **Natalie Twine[2]**, Laurence Wilson[1], Aidan R. O'Brien[1], Piotr Szul[2], Robert Dunne[2], Denis C. Bauer[1]

[1] Transformational Bioinformatics Team CSIRO
[2] Data61 CSIRO

Genome-wide-association studies (GWAS) nowadays often apply Random-Forest for its capability to consider the interactions between genes. Random-Forest is an ensemble technique that adds randomness to Decision Trees, which are individual predictive machine learning models that capture interaction between genomic loci (features). This randomness allows the evaluation of a larger solution space for associated loci in GWAS-style analyses.

One of the important parameters in the Random-Forest is the number of features to be evaluated at each node of each tree (mtry). This parameter directly controls the randomness in the model and substantially affects the performance by potentially limiting the exhaustive exploration of the solution space. This is especially crucial for multi-gene diseases, where sets of features may only incrementally obtain strong disease-association (deep trees) and thereby initially compete with individual features of moderate association (shallow trees). There have been efforts in the literature to find the optimal value of mtry. However, the optimal value highly depends on the dataset and its characteristics.

In our work, we propose a method in which the value of mtry varies during the training process. Thus, not all trees are built using the same mtry allowing the creation of trees with diverse depths. The ensemble hence captures the strongest individual but importantly also sets of features associated with the disease. Furthermore, we evaluate changing the value of mtry at the node level, which allows an even more comprehensive search of the solution space. We assess our approach on Bone Mineral Density (BMD) case/control datasets.

KEY WORDS

GWAS, Random-Forest

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Behnaz Khavari	PhD Student	University of Newcastle	NSW
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Michael Knapp	Senior Lecturer in Biological Anthropology	University of Otago	New Zealand
Joey Lai	Genomics Facility Manager	Westmead Institute	NSW
Michael Lardelli	Associate Professor In Genetics	The University of Adelaide	SA
Hyun Jae Lee	Research Officer	QIMR Berghofer Medical Research Institute	QLD
Ruby Lin	Project Manager	Westmead Institute	NSW

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NAME	POSITION	ORGANISATION	STATE/ COUNTRY
Tian Lin	Research Assistant	University of Queensland	QLD
Ryan Lister	Lab head	University of Western Australia	WA
Huan Liu	Deputy Director	BGI China	China
Limiao Liu	BD Manager	GENEWIZ	China
Ville-Petteri Makinen	EMBL Australia Group Leader	SAHMRI	SA
Courtney Male	Sales Specialist	Bio-strategy Pty Ltd	VIC
Vikki Marshall	Project Manager	Alfred Hospital	VIC
Michael McDonald	Group Leader	Monash University	VIC
Leanne McGrath	Account Manager	AGRF	VIC
Hilary Miller	Application Scientist	Biomatters Ltd	New Zealand
James Miller	Business Development Manager	Millennium Science	VIC
Keith Miller	Regional Sales Manager	Integrated Dna Technology	NSW
Sujanna Mondal	PhD Student	Childrens Cancer Institute Australia	NSW
Sally Mortlock	Postdoctoral Researcher	The Institute for Molecular Bioscience	QLD
Allan Motyer	Postdoc	University Of Melbourne	VIC
Ludovic Orlando	Research Director	University of Toulouse	France
Hardip Patel	Research Fellow	Australian National University	ACT
James Paterson	Postdoctoral Fellow	Flinders University	SA
Katherine Pillman	Bioinformatics Research Fellow	University of South Australia	SA
Gareth Price	Head of Computational Biology	Queensland Facility for Advanced Bioinformatics	QLD
Michael Rhodes	Sr. Dir. Advanced Sequencing Applications	Nanostring Technologies	United States
Michael Ricos	Senior Scientist	University of South Australia	SA
Michael Roach	Postdoctoral	Australian Wine Research Institute	SA
Steven Roberts	Account Manager	BD Biosciences	WA
Akzam Saidin	PhD Student	University of South Australia	SA
Jenny Saleeba	Senior Lecturer	University of Sydney	NSW
Juan Carlos Sanchez Ferrero	Bioinformatician	University Of Adelaide	SA
Andreas Schreiber	Head of Bioinformatics	SA Pathology	SA
Martin Smith	Head of Genomic Technologies	Garvan Institute	NSW
Renee Smith	Research Associate	Flinders University	SA
Adam Smith	Applications Specialist	PerkinElmer	VIC
Marlon Stoeckius	Senior Research Scientist	New York Genome Center	United States
Stephanie Sun	Marketing Manager	BGI Australia	QLD

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NAME	POSITION	ORGANISATION	STATE/ COUNTRY
Eileen Tan	Regional Marketing Manager	Twist Bioscience	Singapore
Peri Tobias	Postdoctoral Research Associate	University of Sydney	NSW
Richard Tothill	Group Leader	The University of Melbourne	VIC
Peter Tsai	PhD Student	University of Auckland	New Zealand
Shoji Tsuji	Professor	The University of Tokyo Hospital	Japan
Taru Tukiainen	Academy Research Fellow	Institute for Molecular Medicine Finland	Finald
Natalie Twine	Postdoctoral Fellow	CSIRO	NSW
Gene Tyson	Deputy Director	The University of Queensland	QLD
Mark Van der Hoek	Genomics Fellow	SAHMRI	SA
Carola Venturini	Postdoctoral Scientist	The Westmead Institute For Medical Research	NSW
Nic Waddell	Group Leader	QIMR Berghofer Medical Research Institute	QLD
Xiaochuan Wang	Senior Research Officer	Cancer Council Victoria	VIC
Yayu Wang	Associate Researcher	BGI China	China
Josh Warburton	Business Manager	Decode Science	VIC
Trent Warburton	Director	Trendbio Pty Ltd	VIC
Kyria Webster	Technical Specialist	Genesearch	QLD
Adam Werner	Technical Sales Representitve	Integrated DNA Technologies	VIC
Laura Weyrich	Future Fellow	The University of Adelaide	SA
Trystan Whang	Director	Macrogen Oceania	NSW
Kevin White	Professor	The University of Chicago	United States
Deborah White	Director of Cancer Research	SAHMRI	SA
Liam Williams	Auckland Genomics	University of Auckland	New Zealand
Naomi Wray	Professor	The University of Queensland	QLD
Sunny Wu	PhD Student	Garvan Institute of Medical Research	NSW
Sue Yap	General Manager	New England Biolabs Inc	Singapore
Emma Yoo	Sales Manager	Macrogen Oceania	NSW

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