

2014/15

# BioInfoSummer

Monash University, Caulfield Campus  
1-5 December 2014





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Monash University, 1–5 December 2014

# 2014-15

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# Introduction

**B**ioInfoSummer is one of the Australian Mathematical Sciences Institute's (AMSI) major annual educational and outreach programs. Over 200 researchers and students gathered at BioInfoSummer 2014 to increase their understanding and skill levels in bioinformatics with the help of an outstanding array of Australian and international keynote speakers.

Bioinformatics is an exciting discipline analysing and simulating both the structures and processes of biological systems.

Biological experiments can generate large amounts of data, bioinformatics is about acquiring, managing, analysing and understanding that data. It is a truly interdisciplinary field that uses mathematics, statistics and information technology to analyse large and complex biological datasets.

Bioinformatics plays a crucial role in our understanding of genes and cellular structure. In health and medical fields, bioinformatics enables advances in areas such as drug discovery, diagnostics,

and disease management. Bioinformatics is also important in agriculture allowing the identification of unique adaptations, desirable properties and differences between populations.

BioInfoSummer 2014 was hosted by MAXIMA at Monash University and was funded jointly by the Department of Education and Training, the Australian Bioinformatics Network and the Australian Mathematical Sciences Institute, with support from EMBL Australia, Bioplatforms Australia and CSIRO.

**“I congratulate the Australian Mathematical Sciences Institute (AMSI) on running BioInfoSummer to assist researchers across a range of disciplines to develop key statistical and mathematical skills that will enable them to pursue innovation.”**

Senator the Hon Scott Ryan  
Parliamentary Secretary to the Minister for Education



**A**t the opening Professor Geoff Prince, AMSI Director, welcomed participants to the conference. Professor Kate Smith-Miles and the Vice-Provost Professor Ian Smith, Monash University, outlined the importance of gathering researchers of all stages of their career to have the opportunity to network and learn from each other. Then BioInfoSummer 2014 was officially opened by Senator the Hon Scott Ryan, Parliamentary Secretary to the Minister for Education, who inspired attendees by highlighting the importance of the discipline:

*"...Data collection and processing is one of the most talked about topics in the sciences at the moment.*

*We live in a world where we can record more information than ever before.*

*But this information only has a value if we develop the ability to process and analyse it.*

*This is as true for the biological sciences, as it is for all other areas of science and technology.*

*The application of advanced mathematical, statistical and computational techniques to discover, analyse and simulate the structures and processes*

*of biological systems will shape our future.*

*In health and medical fields, the analysis enabled by bioinformatics has led to advances in areas such as genetics, drug discovery, diagnostics, and disease management.*

*And it is through this cutting edge research that scientists and business will develop new products and services, new health treatments and new technologies.*

*As a result we will live longer, more productive lives.*

*Bioinformatics also plays an important part in detecting, monitoring and responding to the immediate challenge of managing emerging infectious disease.*

*This year, we have witnessed the spread of the dangerous Ebola disease through a number of African countries and beyond.*

*I was pleased to learn that the bioinformatics community responded swiftly to the Ebola outbreak in Sierra Leone through the development of a new Ebola genome browser.*

*This will support global efforts to develop cures and treatments, to help forecast Ebola's future course and to assess the impact of various management strategies.*

*BioInfoSummer – bringing together students, researchers and professionals to share state-of-the-art technologies and learn about the use of mathematics and computational science in biological contexts – is clearly an investment in the future of science and innovation in Australia."*

*Communicating with peers, academics and the general public – all while getting paid..."*

Full speech: [www.bis14.amsi.org.au](http://www.bis14.amsi.org.au)

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## Monday 1 December

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### Introduction to molecular biosciences and bioinformatics

An introduction to DNA and genomics, gene expression and genetic regulation, protein structure and function, and cell signaling pathways.

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## Tuesday 2 December

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### Next-generation DNA sequencing and sequence evolution

Analysis of next generation sequencing data, including RNA seq data, and applications to the study of diseases with a focus on molecular evolution and phylogenetics.

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## Wednesday 3 December

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### High-throughput technology and omics data analysis

Looking at microarray and proteomic technologies, covering a number of topics from experiment design, statistical analysis of omics data, to network development using various omics datasets.

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## Thursday 4 December

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### Methods in bioinformatics

A focus on mathematical, statistical and computing methods that have been used widely in bioinformatics. Topics included networks and machine learning, Bayesian methods, EM algorithm and application, and hidden Markov models.

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## Friday 5 December

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### Systems Biology

An introduction to methodologies for network construction, model development and network inference, introducing various deterministic, stochastic and multi-scale mathematical models at the pathway, tissue or organ level.

*“The thing that I enjoyed most was the opportunity to talk to researchers in different fields and improve my knowledge.”*

Hamdan Awan, University of New South Wales

Speaker	Talk	Organisation
Dr Nathan Watson-Haigh	<i>From Microarrays to Co-Expression Networks</i>	Australian Centre for Plant Functional Genomics
Associate Professor Peter Pivonka	<i>Application of disease systems analysis in osteoporosis</i>	Australian Institute for Musculoskeletal Science
Dr Annette McGrath	<i>RNA-seq differential expression</i>	CSIRO
Dr David Lovell	<i>The Australian Bioinformatics Network</i>	
Dr Milica Ng	<i>Careers Panel</i>	CSL Limited
Dr Janusz Dutkowski	<i>Overview of network methods in bioinformatics</i>	Data4Cure
Dr Daniel Zerbino	<i>Ensembl Regulation: surveying the markers of cell differentiation across the data matrix</i>	European Molecular Biology Laboratory-European Bioinformatics Institute
Dr Mark Cowley	<i>Using Next Generation Sequencing to uncover tumour heterogeneity, tumour evolution, and early cancer detection</i>	Gavin Institute
Monther Alhamdoosh	<i>AMSI Internship: A Comprehensive Evaluation of Pathway Analysis Tools</i>	La Trobe University
Professor Phoebe Chen	<i>Pattern Discovery for Biomedical Applications using Bioinformatics Technologies</i>	
Dr Steve Androulakis	<i>Introduction to Python</i>	Monash University
Dr Stuart Archer	<i>Introduction to Python</i>	
Associate Professor Sureshkumar Balasubramanian	<i>Introduction of biology to maths 2</i>	
Dr Richard Beare	<i>Introduction to image processing and quantitative analysis using ImageJ/Fiji &amp; Automating image analysis using Python scripting</i>	
Professor Roger Daly	<i>Identification of novel treatment strategies for human cancers through integrative phosphoproteomics and kinomics</i>	
Dr Jonathan Keith	<i>Bayesian methods in bioinformatics</i>	

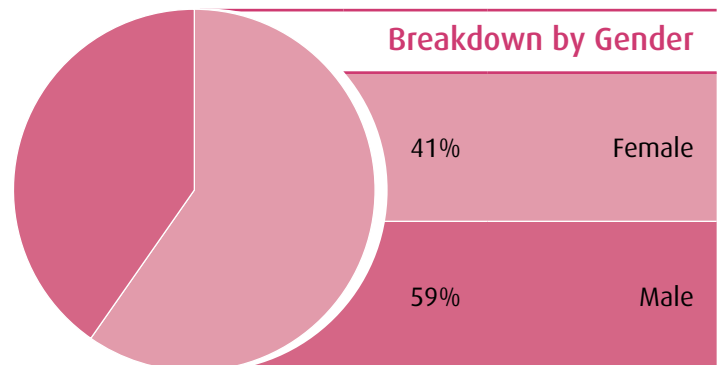


<b>Speaker</b>	<b>Talk</b>	<b>Organisation</b>
Dr Simon Michnowicz	<i>Introduction to Python</i>	Monash University
Dr Saskia Reibe-Pal	<i>Careers Panel</i>	
Professor Falk Schreiber	<i>Modelling and Visualising Metabolism</i>	
Dr Keith Schulze	<i>Introduction to image processing and quantitative analysis using ImageJ/Fiji &amp; Automating image analysis using Python scripting</i>	
Professor Kate Smith-Miles	<i>A few equations that changed biology</i>	
Dr Tianhai Tian	<i>Stochastic modelling and simulation of genetic regulatory networks</i>	
Dr Alicia Oshlack	<i>Transitioning from mathematics to biology: what you need to know</i>	Murdoch Childrens Research Institute
Professor Edmund Crampin	<i>Introduction to Systems Biology</i>	The University of Melbourne
Professor Marc Wilkins	<i>Proteomics and quantitative mass spectrometry</i>	The University of New South Wales
Professor Mark Ragan	<i>Phylogenetics without multiple sequence alignment</i>	The University of Queensland
Associate Professor Jean Yee Yang	<i>Statistics in bioinformatics</i>	The University of Sydney
Professor Chris Overall	<i>Forward perspectives in bioinformatics</i>	University of British Columbia
Dr Barbara Holland	<i>Introduction to model-based methods of phylogenetic inference</i>	University of Tasmania
Professor David Page	<i>Machine Learning from Genomic and Clinical Data</i>	University of Wisconsin-Madison
Simon Gladman	<i>LAB: The Genomics Virtual Laboratory and what it can do for you &amp; hands-on computer lab</i>	Victorian Bioinformatics Consortium
Associate Professor Andrew Lonie	<i>LAB: The Genomics Virtual Laboratory and what it can do for you &amp; hands-on computer lab</i>	Victorian Life Sciences Computation Initiative
Dr Clare Sloggett	<i>LAB: The Genomics Virtual Laboratory and what it can do for you &amp; hands-on computer lab</i>	
Dr Wei Shi	<i>LAB: A Bioconductor R pipeline for the accurate and efficient analysis of RNA-seq data</i>	Walter And Eliza Hall Institute
Professor Gordon Smyth	<i>RNA-seq: from reads to genes to pathways</i>	

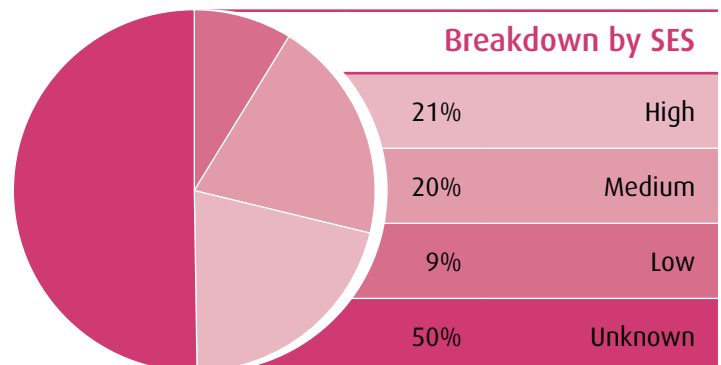
## Enrolments by Institution

Australian Bureau of Statistics	1
Australian Centre For Plant Functional Genomics	1
Australian National University	2
Australian Regenerative Medicine Institute	1
Baker IDI Heart and Diabetes Institute	1
Baylor College of Medicine	1
Burnet Institute	1
CSIRO	4
CSL Limited	1
Curtin University of Technology	1
Data4Cure	1
Deakin University	3
Defence Science and Technology Organisation	2
European Molecular Biology Laboratory-European Bioinformatics Institute (Embl-Ebi)	1
Federal University Dutse	1
Flinders University	1
Garvan Institute	1
Institute for Molecular Bioscience	1
Interdisciplinary Centre for Bioinformatics	1
Latrobe University	11
Ludwig Cancer Research	1
Macquarie University Sydney	3
MAXIMA	3
Monash University	63
Murdoch Childrens Research Institute	1
Other	2
Peter Doherty Institute for Infection and Immunity	1
Peter Mac	1
Queensland University Technology	1
RMIT University	15
Stobor Pty Ltd	1
Swinburne University of Technology	1
The Institute of Structural and Molecular Biology (ISMB)	1
The University of British Columbia	1
The University of Melbourne	36
The University of Queensland	3
University of Adelaide	6
University of Canterbury	1
University of Exeter	1
University of New South Wales	3
University of Newcastle	5
University of Sydney	3
University of Tasmania	3
University of Western Australia	3
University of Wisconsin-Madison	1
Victorian Life Sciences Computation Centre	2
Walter and Eliza Hall Institute of Medical Research	5
<b>Total</b>	<b>204</b>

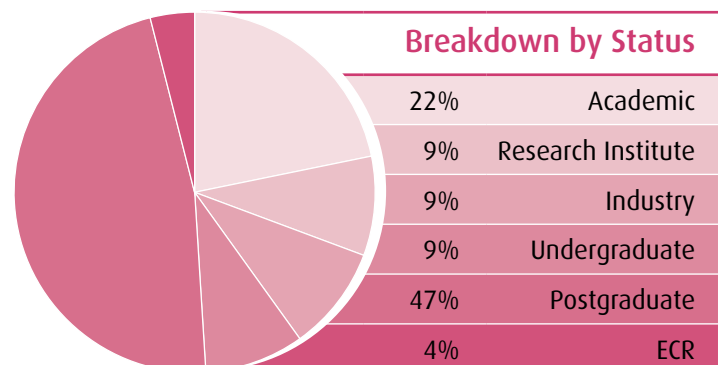
## Breakdown by Gender



## Breakdown by SES



## Breakdown by Status



Number of participants identifying as Aboriginal or Torres Strait Islander: **1**

# TOO HOT... TOO COLD...

STATISTICIANS HELPING TAILOR YOUR DRUG TREATMENT TO GET IT... JUST RIGHT.



**Andrew Pfeiffer**

**A**ndrew Pfeiffer's studies will help health professionals tailor prevention programs to each of their patient's unique genetic makeup. In addition, if a patient becomes ill, the same information can be used to select treatments most likely to be effective and least likely to cause adverse reactions.

In his second year of a master's in statistics Andrew is researching genome-wide association studies. The impact on medical care from genome-wide association studies could be substantial. Andrew's research area is laying the groundwork for the era of personalised medicine, in which the current one size-fits-all approach to medical care will give way to more customised strategies.

In the future, after improvements are made in the cost and efficiency of genome-wide scans and other innovative technologies, health professionals will be able to use the tools Andrew is working on to provide patients with individualised information about their risks of developing certain diseases.

*"Attending BioInfoSummer 2014 reminded me that I still have much to learn (don't all researchers?), but I am much more confident now regarding how I can teach myself more about the various aspects of bioinformatics now and into the future. Having an annual symposium allows attendees to become aware of and fascinated by the exciting and high-paced field of bioinformatics – I really enjoy the mix of mathematics, statistics, computer science and biology presented,"* Andrew says.

BioInfoSummer 2014 was Andrew's second BioInfoSummer. In 2013 Andrew remembers feeling quite clueless during

some of the presentations, but he says that being amongst other young researchers he realised this was the norm. *"BioInfoSummer 2013 taught me that as a researcher it is my job to look up the terminology and piece together my own understanding of whatever field I am interested in. It was a great introduction to research life,"* says Andrew.

As someone with a background in mathematics and statistics, Andrew explains that AMSI's programs motivate him to learn more about computer science and biology. *"BioInfoSummer confirms the importance of good communication between "wet-lab" researchers and "dry-lab" researchers (statisticians, software developers and engineers and mathematicians) to output high quality research for the benefit of all,"* Andrew says.

*"When I started my masters, I hadn't studied any biology in almost ten years,"* Andrew says, *"That had to change quickly, my research studies the statistical association between biological pathways and diseases."*

The practical sessions, explains Andrew, are fantastic opportunities to learn new skills and put them into practice. *"And the careers panel, consisting of researchers from government agencies, universities and private industry, allow us to have a discussion about the state of bioinformatics in Australia and abroad,"* Andrew says.

*"I am aiming to find employment for next year. It would be amazing if I could work in bioinformatics!"* concludes Andrew.

## Poster Competition

On Thursday 4 December attendees got the chance to showcase their research with poster presentations.

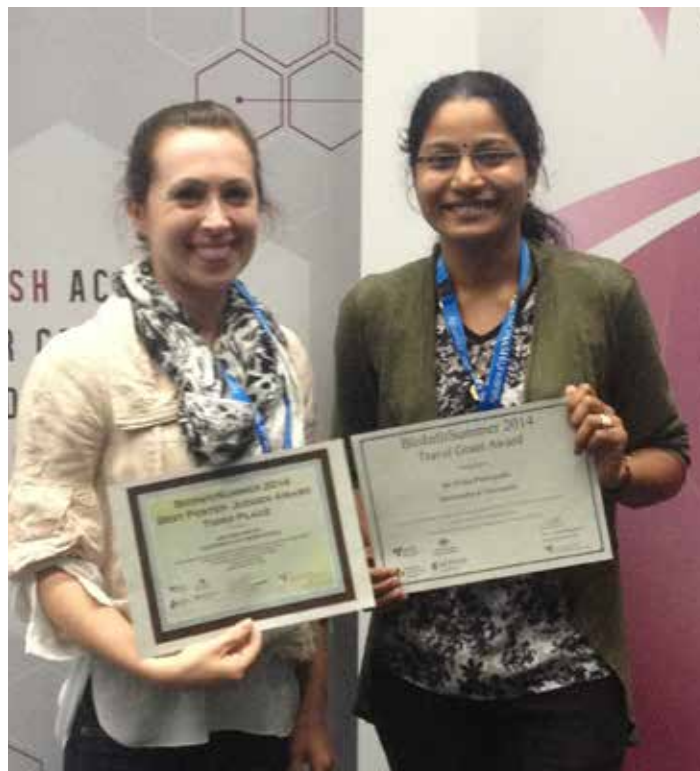
### Judges Prizes

- 1st place:** Dane Vassiliadis, University of Melbourne
- 2nd place:** Manjula Algama, Monash University
- 3rd place:** Qianqian Wu, Monash University & Haloom Rafahi, Baker IDI Heart & Diabetes Institute  
Heloisa Milioli, University of Newcastle

### People's Choice Award:

Haloom Rafahi, Baker IDI Heart and Diabetes Institute

The poster session also allowed participants to network over drinks and discuss their research.



## Travel Grants

The Australian Bioinformatics Network, EMBL Australia and BioPlatforms Australia annually offer travel support for students to attend the event. This year sixteen students and early career researchers received travel awards:



- Shanika Amarasinghe, The University of Adelaide
- Luis Alberto Arriola-Martinez, The University of Adelaide
- Jaelle Brealey, The University of Queensland
- Reuben Buckley, The University of Adelaide
- Joshua Brzozowski, University of Newcastle
- Sally Louise Crane, University of New South Wales
- Atma Ivancevic, The University of Adelaide
- Helen Maria Jankowski, University of Newcastle
- Mythreye Krishnan, The University of Western Australia
- Andrew Pfeiffer, The University of Adelaide
- Nisha Puthiyedth, University of Newcastle
- Van Nam Hoang, The University of Queensland
- Abaraham Shibesh, Australian Bureau of Statistics
- Sitthichoke Subpaiboonkit, The University of Queensland
- Qi Yang, Flinders University
- Lu Zeng, The University of Adelaide



# Careers Panel

The careers panel is an opportunity for students to find out more about careers in bioinformatics and to engage the wider community. The panel consisted of five people at varying stages of their career, from industry and academia. The panel engaged the audience with lively discussion and answered a barrage of questions, inspiring the next generation with career opportunities in the discipline.



Dr David Lovell, *Bioinformatics & Analytics Leader, CSIRO*



Dr Milica Ng, *Research Scientist, Bioinformatics, CSL Limited*



Dr Janusz Dutkowski, *Founder, Data4Cure USA*



Dr Saskia Reibe-Pal, *Researcher, Monash University*



Professor Chris Overall, *University of British Columbia*





# CABBAGES

have twice as many genes as you. No? Yes... really!

Interview with *Chris Overall*

**C**hris Overall has an impressive lineage; scientifically and culturally. Not only is he Australian, but his Post Doctoral supervisor is a Nobel Laureate (Dr Michael Smith, invented site-directed mutagenesis). AMSI's Stephanie Pradier spoke with Chris this week about his current research (terminomics) and BioInfoSummer 2014 (BIS14).

Chris prefaced our conversation by declaring that he comes to bioinformatics not as a mathematician or a statistician or a computer scientist, but as a user. In fact, he was quite pleased when the Australian Mathematical Sciences Institute (AMSI) approached him to give the opening lecture at BIS14.

"I am not going to talk about the past of bioinformatics, where it has come from, where it is going. I am not qualified to do that," Chris said. "I would like my audience to leave aspiring to be the Steve Jobs — the Apple — of bioinformatics. To design and code easy to navigate, beautiful programs and interfaces that users, like me, want to come back to and use again and again."

He said it is important for the user to be able to "go under the hood" if they need or wish to, but that it shouldn't be necessary.

Chris is a Professor and Canada Research Chair in Protease proteomics and Systems Biology. He works at the Overall Lab — yes, he even has a lab named after him — a proteomics and protein engineering research laboratory.

Set up in the early 90s, the team at Overall Labs (which currently consists of Chris, two research associates, ten post docs and two PhD students) are developing tools to identify targeted ways to combat illnesses. Chris explained to me that medical research aims to understand the molecular pathways

forming the basis of disease so that we can identify targets or their pathways capable of being used to develop drugs or treatments to counteract these, and so treat these diseases. However, every five to ten years we find another layer of complexity.

He broke this down for me: "Humans have 20,135 genes; a cabbage has 41,174; a London double decker bus has 24,000 different parts. So the question is, on the basis of the assumption of one protein per gene, how do we (humans) have so much more complexity than a cabbage if they have twice as many genes as we do?"

"Part of the answer lies in the timing of the genes, that is when they turn on and off," Chris said. "The other part is what are called post-translational modifications, or PTMs. After the gene synthesises RNA the RNA then makes a protein — we call the production of new proteins from these parent protein chains protein post-translational modifications and this is what generates millions of forms of proteins in humans and hence generates incredible complexity from which life arises.

"So, post-translational modification looks out how the proteins are mixed and matched. It turns out that there are, on average, five alternate splice forms per protein. Which means that we can have different versions of the same protein depending on how it is made. If we multiply our assumption of one gene, one protein by five we now have 100,000 different proteins. Then there are a further twenty amino acids that make up these proteins, and many of these amino acids can be affected by chemical modifications — giving us even more diversity. In fact, doing the maths takes us up to around five million different targets; much more reasonable for a complex, self-healing, reproducing organism."

Chris's research steps one more rung up the ladder: "I investigate the ends of the proteins," he said.

Enzymes cut through proteins, removing amino acids. And sometimes, missing just four amino acids — from a couple of hundred — can completely change the protein from giving a "go" signal to a "no-go" signal.



"This is what we study at the lab," Chris informed me. "We call it terminomics," he continued, with a jovial tone to his voice. "We have found lots of great examples where unless you know what the ends are, you don't know whether the protein is on or off – whether it is an antagonist or protagonist."

This is where Chris believes a big problem for bioinformatics arises. He gives me an example: "You have a particular protein, you find it in a database and you say to yourself, 'This protein is present, therefore this interaction network must be present and therefore the pathway must be...' But unless you are sure that the ends of the protein are there, those four amino acids, you would be completely wrong. So this is what we study; how molecule functions change depending on how they are cut."

Fairly green to this field of study, I asked Chris if they do the cutting of the proteins with the enzymes to see which amino acids are removed. He laughed as he replied, "That's old school. I started my PhD adding the enzyme to the protein to see where it gets cut and then characterising it. We look at whole tissues now and determine all the cut ends at once," Chris finished with a smirk.

The computer programs available now are simple; they don't take into account the deeper interactions. In order for this to happen, Chris said that bioinformaticians need to work closely with biologists, "We have always had one or two coders, and so we have developed a suite of programs specifically designed to analyse our proteomics data sets and to make these readily accessible for knowledge translation. Our TopFIND knowledge base has just been updated to v3beta with over 170,000 termini instantly accessible with protein maps, interaction paths, original references and other tools for analysis. It is by no means perfect and Steve Jobs would still most likely turn in his hard drive, but we are continually refining it to make it easier for the user.

"Don't sit in a silo. As a bioinformatician get yourself embedded in a good biological lab and then you'll be able to see the experiments and understand first hand what sort of questions need to be asked of the data."

When Chris said this I immediately recalled hearing Professor Kate Smith-Miles say something very similar: "When you are collaborating with biologists you have to understand their language first and understand what their problem is before you can even get to the point of applying any mathematics."

Kate went on to explain that over her career she has spent a great deal of time learning enough about biology to ensure she can have conversations with a variety of biologists. She said it is the essence of the problem that you need to understand in order to be able to describe it as a mathematical problem.

"Bioinformatics is evolving rapidly," Chris said. "Currently we have an amazing capability and access to incredible data sets. What I hope to gain from BioInfoSummer is insight into this evolution, to talk to and learn from my peers. And I hope that my experience as a user can help contribute to the depth and diversity of modern bioscience and systems biology."

Chris said that it is great that AMSI organises events such as these that build platforms and networks for interdisciplinary collaboration. "Collaboration is key in medical research. We need to have collaborative networks as complex as the proteins that make us up. And in the end," Chris concluded, "... you hope, with a little intuition, some clever experiments and good bioinformatics we will be able to map pathways and find targets that combat infection and disease."

Since 2012 EMBL Australia and BioPlatforms Australia have supported AMSI internship placements linked to BioInfoSummer. The program builds research-industry partnerships and establishes long-term industry-research partnerships in bioinformatics.

Under the guidance of an experienced researcher, postgraduate students complete a 4-5 month project with an Australian business. The internships provide access to the vast research expertise in Australia's universities and bring new perspectives and the latest knowledge to a research challenge.

Students awarded internship placements in 2013 presented their projects at BioInfoSummer 2014.



*“Bioinformatics is such a broad topic... and was amazing to hear from speakers that our generation can make the difference!”*

Nisha Puthiyedth, **University of Newcastle**

## PROJECT:

# Comprehensive Evaluation of Pathway Analysis Tools



### Intern:

Monther Alhamdoosh,  
La Trobe University

### Mentor:

Dr Matt Ritchie, The Walter  
& Eliza Hall Institute

### Industry Partner:

CSL Australia

CSL has recently developed its own bioinformatics pipeline for next-generation sequencing (NGS) data analysis. The pipeline enables researchers to identify differentially expressed genes (DEGs) from RNA-seq data generated from in vitro and in vivo models to better understand the mechanisms of action (MOA) of pre-clinical and clinical drugs.

The aim of this project was to extend the analysis pipeline to summarise these results at the level of molecular pathways and signalling networks. A comprehensive evaluation of two leading commercial software tools for pathway analysis was conducted in order to assess their ability to efficiently and accurately infer the pathways and biological processes that are perturbed by a list of DEGs.

The project has provided CSL with a clear assessment of the available commercial and open source pathway analysis software that has enabled them to make an informed decision on how best to expand their bioinformatics capabilities in order to aid their current biomarker discovery processes and improve their understanding of MOA of their drug targets and candidates.





“BioInfoSummer gave me a great introduction to what the field of bioinformatics provides, and the numerous opportunities that it offers”

Phillip Luong, Monash University

“The atmosphere at BioInfoSummer was great. To be surrounded by people working towards a shared goal reinforces my passion for science. And, as a researcher, it is great to bounce ideas off others trying to bridge the same gap: incorporating informatics into biology.”

Ellen Fortini, Harry Perkins Institute of Medical Research

“It was a steep learning experience for me about bioinformatics methods and some of the underlying biology, I learned a lot about next generation sequencing and differential expression. I also found the practical sessions very useful.”

Sinead English, University of Oxford

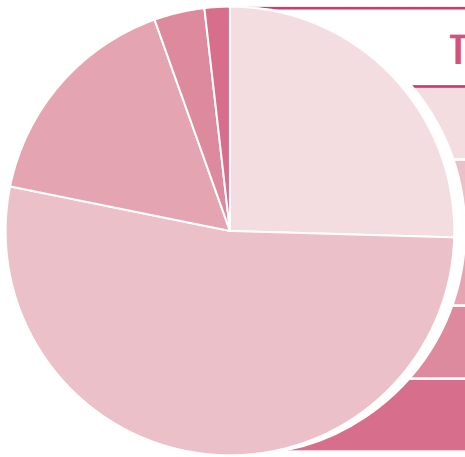
“It was a privilege for me to attend BioInfoSummer2014 at the early stages of my PhD. I was able to present my first poster in the PhD studies there too. The ability to engage and share experiences with the leading figures of my academic area was the most important part of this conference for me.”

Shanika Amarasinghe, University of Adelaide





## The Content Presented Was Relevant To My Needs



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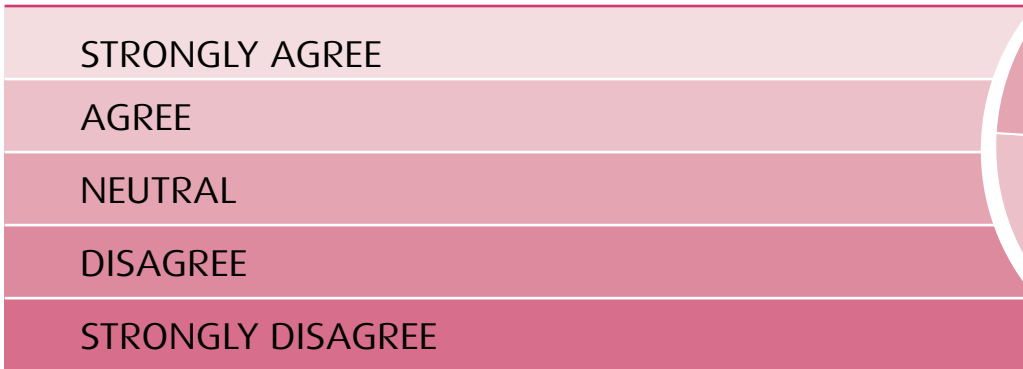
AGREE

NEUTRAL

DISAGREE

STRONGLY DISAGREE

## I Found The Social Events A Good Opportunity To Network



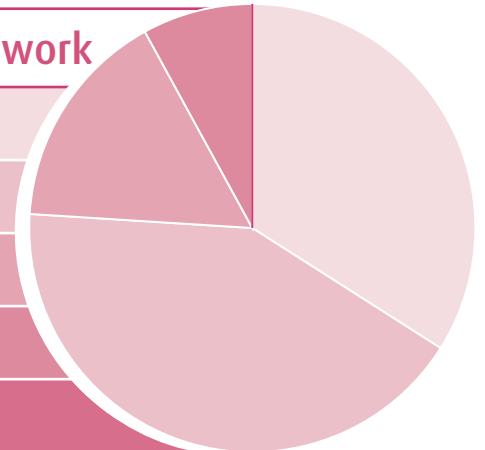
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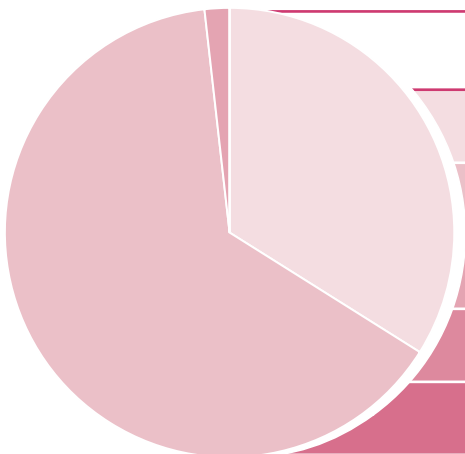
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## The Presentations Were Professional & Engaging



STRONGLY AGREE

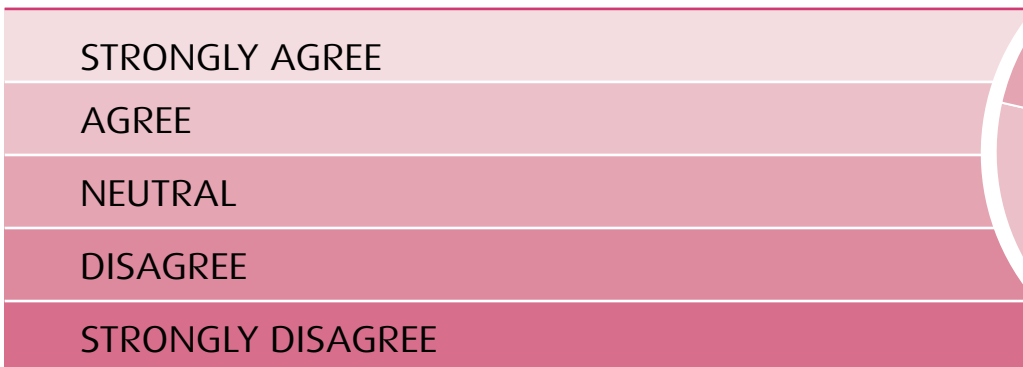
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## I Would Recommend Bioinfosummer



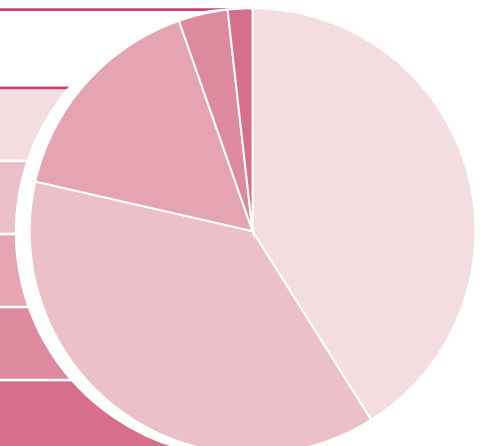
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**T**erry Speed is the head of Bioinformatics at the Walter and Eliza Hall Institute of Medical Research. He is one of Australia's eminent statisticians, a fellow of the Royal Society and 2013 recipient of the PM's Prize for Science are among his many accolades. We spoke with Terry about his research, passions and women in STEM.

## Can you tell us about a research problem that you're currently excited about?

Well, I get my excitement from both the science and the statistics that I need to address the science. One of the areas that I'm really interested in these days is called epigenetics. Epi is the Latin prefix – above, on, over.. – epigenetics is really on top of the DNA, giving guidance to an organism to start doing things with the DNA in a cell: reading (transcribing) the DNA, and converting (translating) it to proteins. In some sense, it's going one step behind what we see. We sometimes see it suggested that DNA is the whole story, but of course there's a story behind the story, and much of the interest in science is sort of peeling back the layers. So for epigenetics, we typically think in terms of gene regulation – governing what genes go on and what genes go off, and when they go on and when they go off. It's pretty complicated because we have many important times in the life of an organism and we have many sorts of cells in the body of a complex organism like us. So it's almost an infinitely complex mosaic to understand. Doing so involves lots of challenging statistical problems and it's fun biology as well. I probably should say why. Consider the impact of diet. For example, if you feed a worker bee royal jelly you convert it into a queen bee. Diet alone changes the way in which the genes are expressed so that although it was originally sterile, it becomes fertile, it's able to lay eggs – some pretty dramatic change just from eating some jelly. There are many examples like that.

## How are you using statistics to make sense of epigenetic data?

One of the topics that I study is methylation, which is a modification of DNA that plays an important role in expression of genes. It can happen across the entire genome, and from the point of view of an observer you're interested in finding out exactly where it's been modified, and how differences of these modifications may play a role. So there's a general

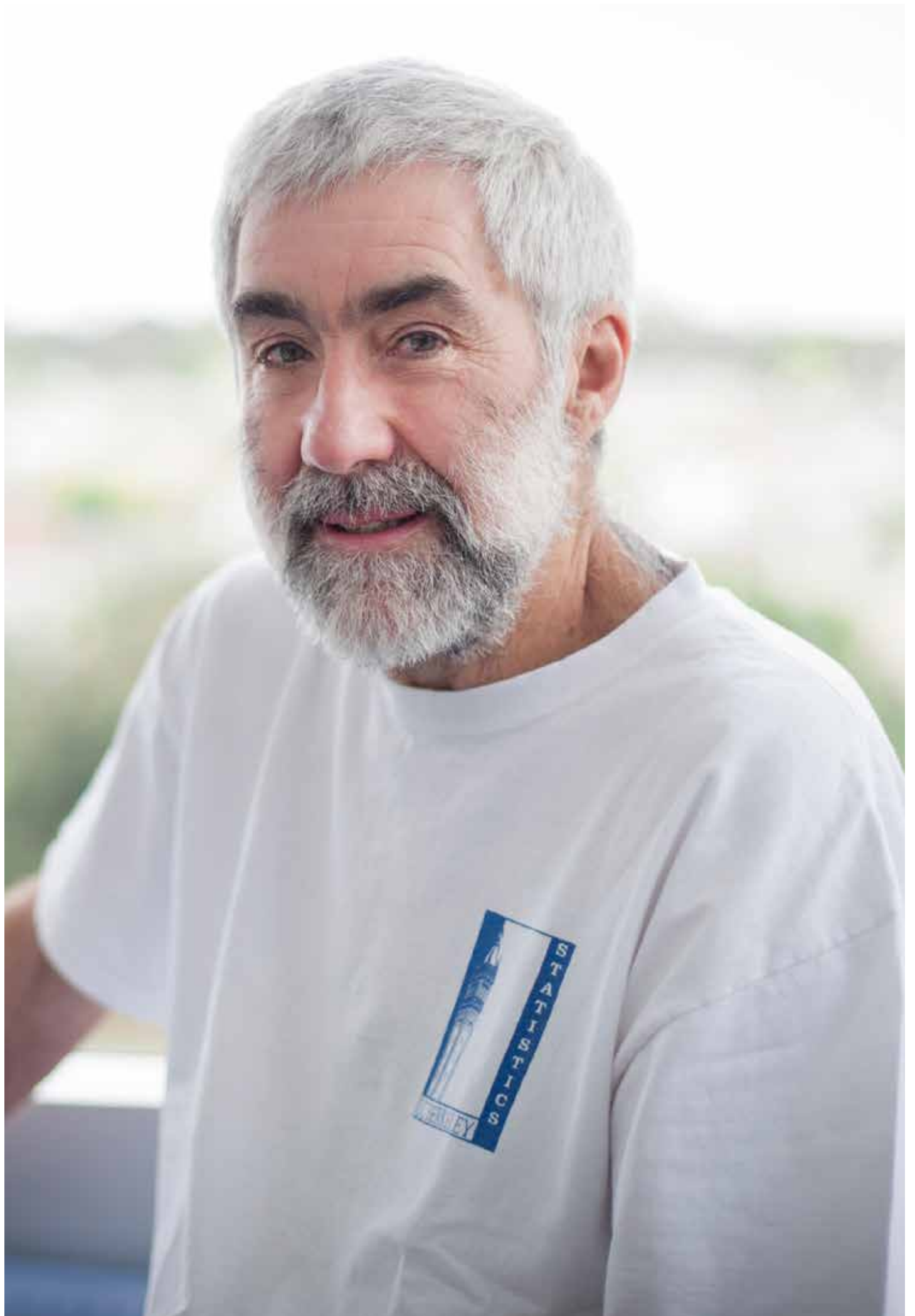
topic of the identification of differentially methylated regions between two sorts of cells. It's a bit like differentially expressed genes, but as I said, one step behind that. There are lots of challenging issues here. There's serial correlation in the methylation marks along the genome. That's quite complex and it mixes in with a lot of genome structure. So, just the simple task of finding which regions are differentially methylated in cell type 1 compared to cell type 2 – there are already a host of interesting statistical challenges in that.

## How important is collaboration in your research?

I think the short answer is 'very important', even right back when I was doing my PhD, essentially on my own. Of course because a PhD isn't supposed to be collaborative, so I used to say that I was a social mathematician, in the sense that I like talking to a lot of people about my work. I'm not that interested in chugging away on my own in total isolation, I like to interact with people, and in a scientific environment it's a natural thing to do: it's called collaboration. With mathematicians, sometimes they collaborate in the sense that they both contribute to the same problem, but it's a lot easier in medical research because you bring different skills to the same issue. In mathematics, perhaps it's less so – often you have the same sort of skills, and you just carve up the problem in different ways. But anyway, the answer, it's very important to me because I don't like working on my own.

## Mathematicians and statisticians are often depicted as boring number crunchers. How can we address this image problem?

Well I think the short answer is we just have to get out there. We have to be involved, not just be background people but be involved a little bit more up front. Dive in, don't be afraid. When you look at physicists or computer scientists, my impression is they tend to be less shrinking violets than we mathematicians and statisticians. But of course we probably think we worry about the details more than big picture people. I'm not speaking against details but you shouldn't let concern for details inhibit you getting involved in big picture things. Have confidence that we in the statistical and the mathematical world have something to contribute – dive in and then be careful, but don't let your careful nature inhibit you before you dive in.



### As a student, who were your scientific heroes?

I really had one above all others and that was R.A. Fisher, the statistician and geneticist who worked in evolution. I had side heroes (e.g. Norbert Wiener), but Fisher stood out. I did a third year Honours project about his work and I learned a lot about him in my courses. Fisher just stood out. He was known to geneticists as a great geneticist, known to statisticians as a great statistician, known to evolutionary biologists as a great evolutionary biologist. That covered a breadth of my interests as an undergraduate and they're still very close to my major interests now. There were other people – Darwin, the Huxleys – people who are involved in evolution of course were and are also very important to me.

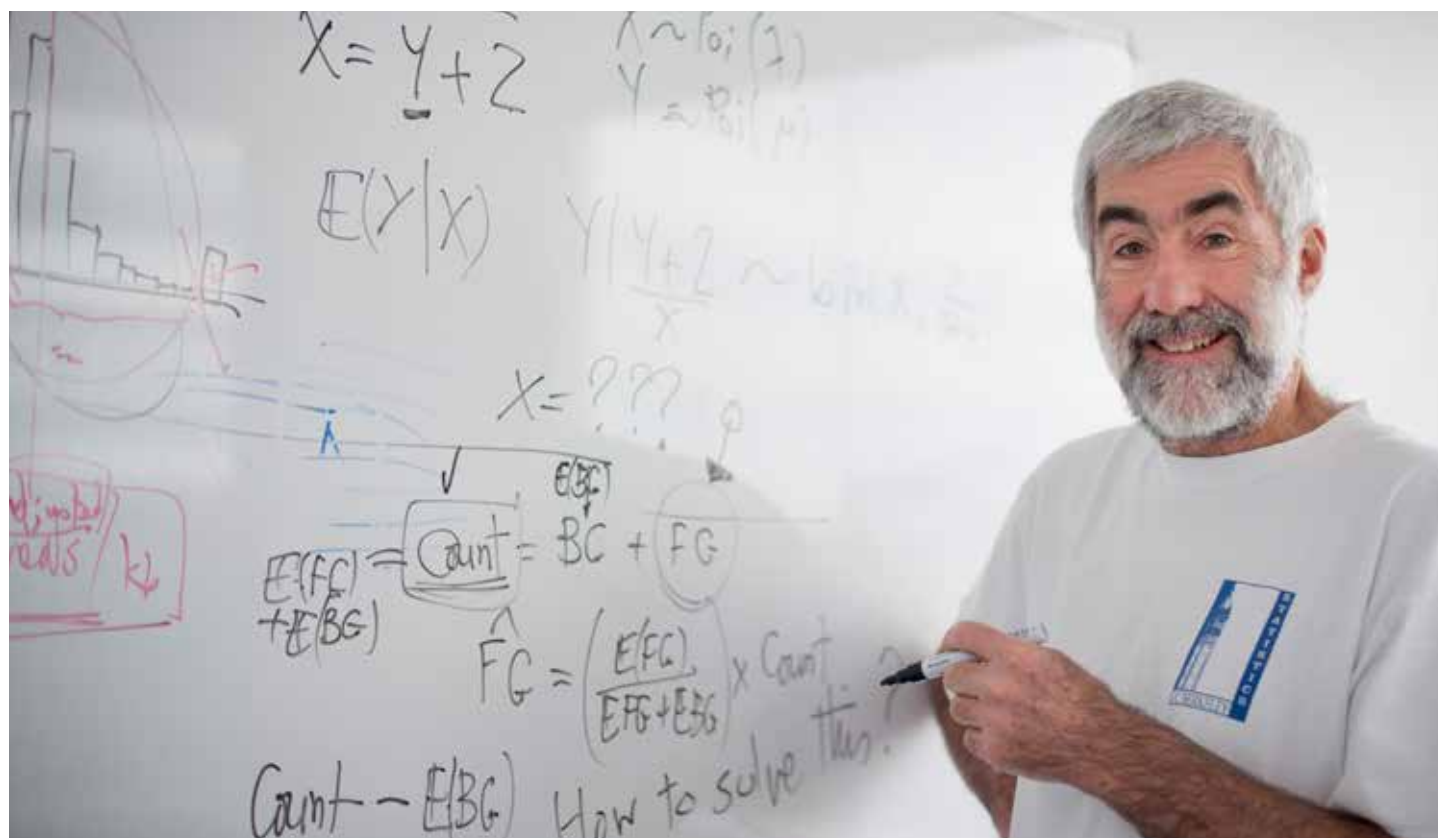
### What attracted you to study mathematics over other sciences?

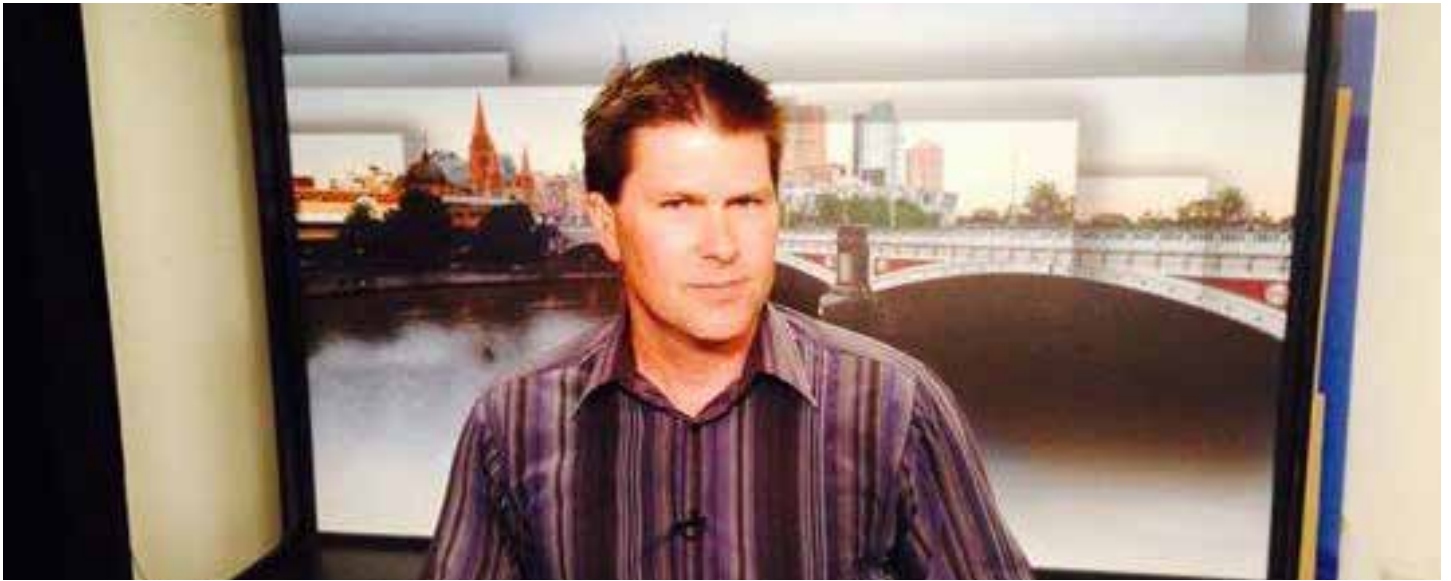
I think it was basically by elimination. I didn't find experimental science to my liking. Experiments are great in theory. In practice, I was frustrated – my experiments never quite worked the way they ought to. I think I have an idea of the world – it's almost a platonic view, that things ought to be perfect. When you do a chemistry or a physics experiment it almost never comes out perfectly. When you do a mathematics experiment, you can get it right, it can come out perfectly. So a desire for perfection, you could say, or if you like a frustration with the imperfections of the real world, led me to this more abstract and more perfect world. Of course

I'm also interested in applying the abstract and perfect stuff to the real world, but that is a rather different activity than doing experiments. I think I'm a person who likes science but is a refugee from experimental science.

### You're a keen advocate for gender equity at the Walter and Eliza Hall Institute of Medical Research. What do you think would improve female participation in maths-related professions?

Being a male, I tend to focus on the things where the males are possibly at fault or at least things men can do. Clearly there are lots of institutional, structural other barriers to women succeeding in so-called STEM discipline – Science, Technology, Engineering and Mathematics. It's not just attitudes of men, but men run the show in most of these areas, so it's a change in the attitudes of men that I think is going to be a major part of changing the situation for women in the STEM disciplines. Being a man and being a supporter of this change, I think I've got a role to play. Clearly women have a role to play as well, but it's not for me to tell them how to live their lives. But if I see men who are being obstructive, who are being conservative or being narrow-minded, who don't look beyond their immediate male counterparts, I'm very happy to speak up and condemn this sort of activity and trying to move towards a more equitable way of, as it were, involving the entire human race in the activities like mathematics and other disciplines.





**B**ioInfoSummer is a highly regarded annual meeting, conference and workshop designed for PhD students and researchers in bioinformatics and related fields. Attendees are principally from Australia, but there is also a small international presence. Its primary function is educational – it provides students and early career researchers with the opportunity to become informed about the scope and direction of Australian research in bioinformatics. It also helps place that research in an international context. It is a unique event in that it is a conference aimed primarily at students, yet is in no way a “mock conference”; it includes presentations of the latest research from scientists at the forefront of the field.

The 2014 meeting was very well attended, with over 200 attendees. One highlight of the 2014 meeting was the keynote address by Professor Chris Overall, Research Chair in Proteinase Proteomics and Systems Biology at the University of British Columbia, Centre for Blood Research. Professor Overall presented outstanding research on protease interactions, and emphasised an important guiding principle for research in bioinformatics, namely that “bioinformatics must be accessible, easy and beautiful to use in order to be useful to the broad community”. Another highlight was the informal welcome speech by Professor Terry Speed, which included important insights into the employment prospects for students of bioinformatics in Australia and abroad.

The organisers received very positive feedback regarding the poster session. Most of the posters were presented by students, and a number of researchers remarked on the high quality of both the students and the work presented. Students appreciated the valuable feedback and suggestions they received from more senior researchers. Assoc. Prof. Barbara Holland commented that this event was also “incredibly fun”; the close quarters layout and provision of food seems to have fostered a particularly interactive and vibrant session.

As in previous years, computer software sessions were well attended and well regarded. Sessions on the Genomics Virtual Laboratory in particular introduced many of the students to a powerful computational infrastructure resource available to Australian researchers. Despite several logistical difficulties encountered during the organisation of these sessions, feedback indicated that they were well worth the effort.

The 2014 meeting continued to enhance the already strong reputation of the BioInfoSummer series as a forum for students and early career researchers in bioinformatics to expand their knowledge base and make valuable connections that will benefit their current and future research.





AMSI wishes to acknowledge the generous donation of time and scientific advice of the following committees, without their contribution this event would not be a success.

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image of parallel telomere quadruple created by Thomas Spletstoesser

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