

**A student's journey—how Bioinformatics
changed my worldview: the many platforms,
data types and analyses I've learnt along the
way.**

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There and back again...

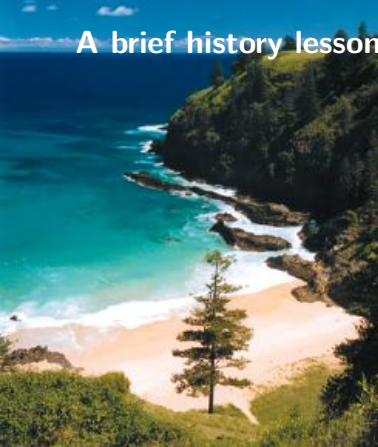


A journey in Bioinformatics

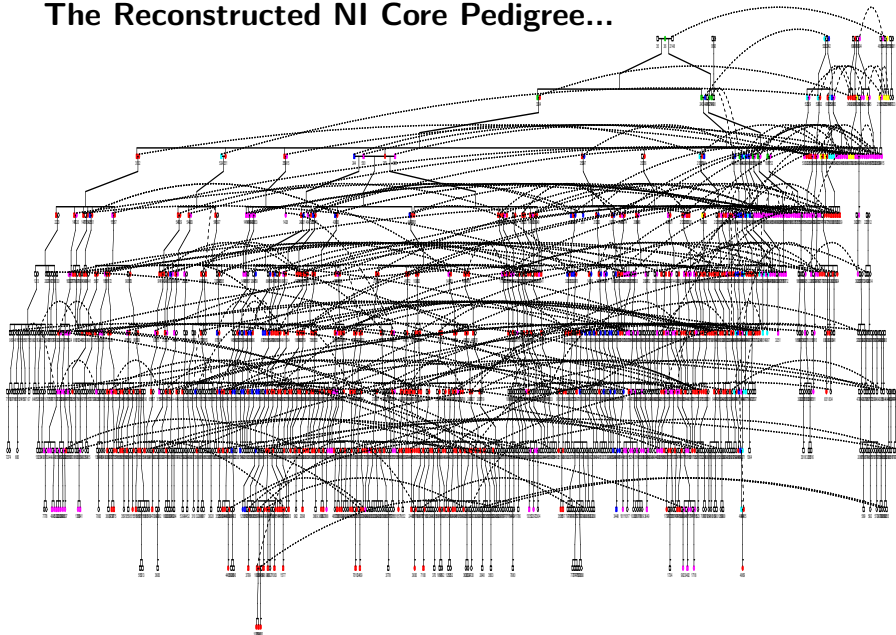
Norfolk Island



A brief history lesson...



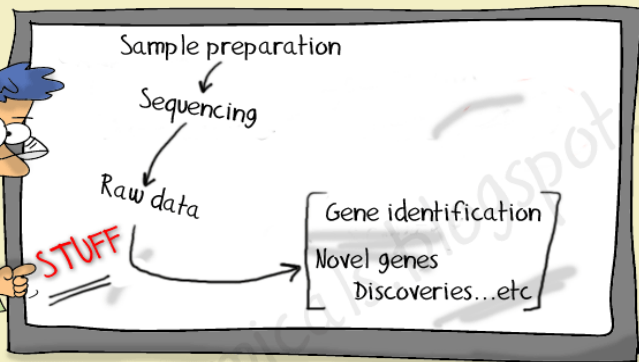
The Reconstructed NI Core Pedigree...



Bioinformatics

What is bioinformatics?

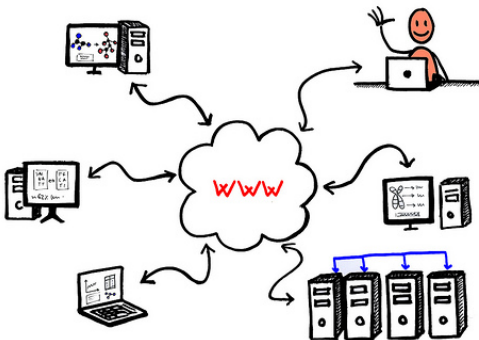
We are
bioinformaticians
thats what we do



What is Bioinformatics?

"the new molecular biology"

Labs are now also in silica...

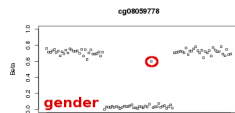
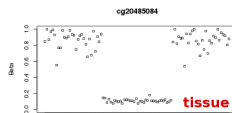
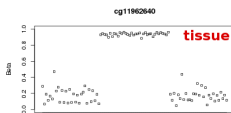
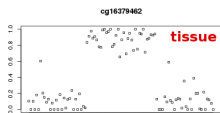
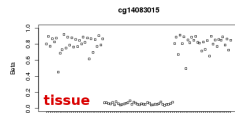
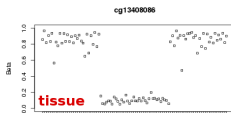
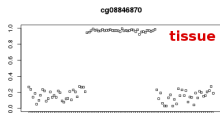
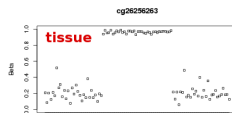
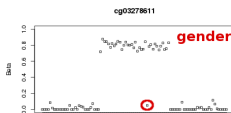
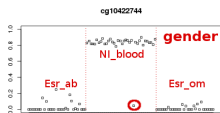


Study Design is Crucial

Need to really think about study design:

- Same old story: Quality in equals Quality out
 - good bioinformatics will never make up for poor lab technique and/or poor study design
- Gender - sex specific methylation patterns
- Origin of sample (i.e. blood, tissue)
 - mixed cell populations
- Case/Control? Paired-samples? Numbers?

Example: Gender and Tissue specific markers



Appropriate QC and Statistical Analysis

Quality Control

- Probe failure
 - always an issue with high-throughput - how do we deal with this?
- Normalisation of data... Yes, but if too stringent you lose information!

Statistical Analysis

- High-throughput Methylation data is 'special'
 - most probes won't hold to normality
- Need to put thought into specific tests based on design of study
 - case/control design will use non-parametric
 - paired samples can get away with more 'popular' stats
- Multiple testing and Significance thresholds?

Important lessons...

Being inquisitive and 'meticulous' pays off...

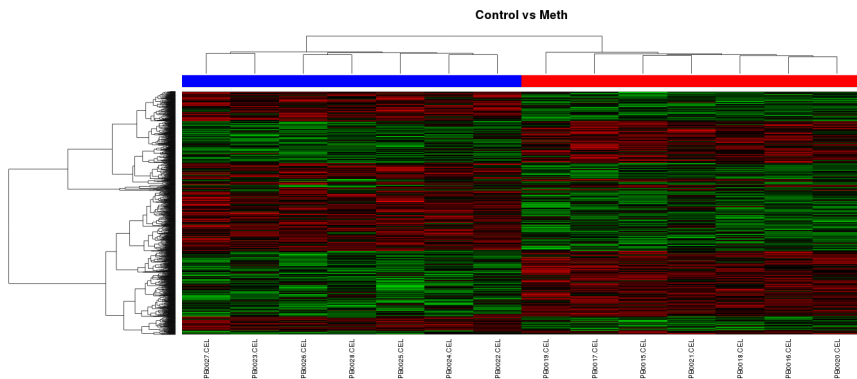
Communication

Collaboration

'Rabbit Holes' are exciting!

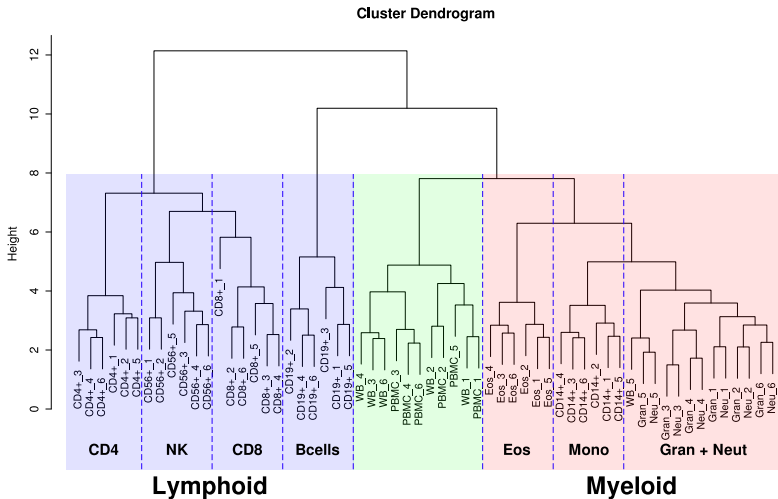
Drugs of Addiction: Methamphetamine

Truly 'multi-omics' approach (mRNA, miRNA and protein)



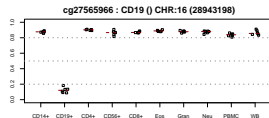
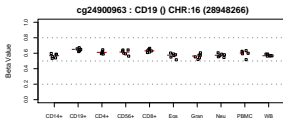
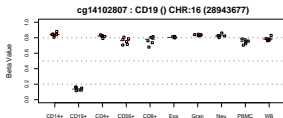
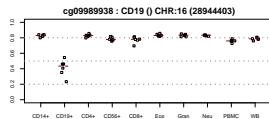
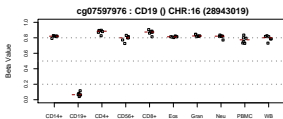
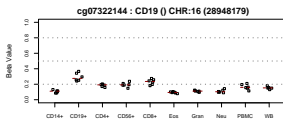
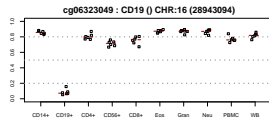
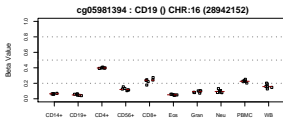
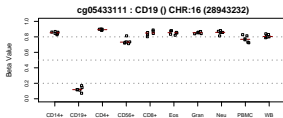
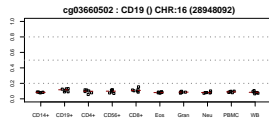
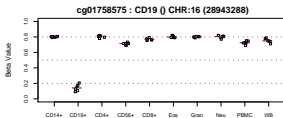
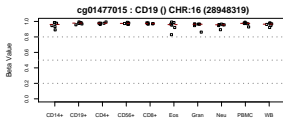
Investigating Methylation Profiles in Blood Cells

Publicly available data set (6 'healthy' males) - cell sorted

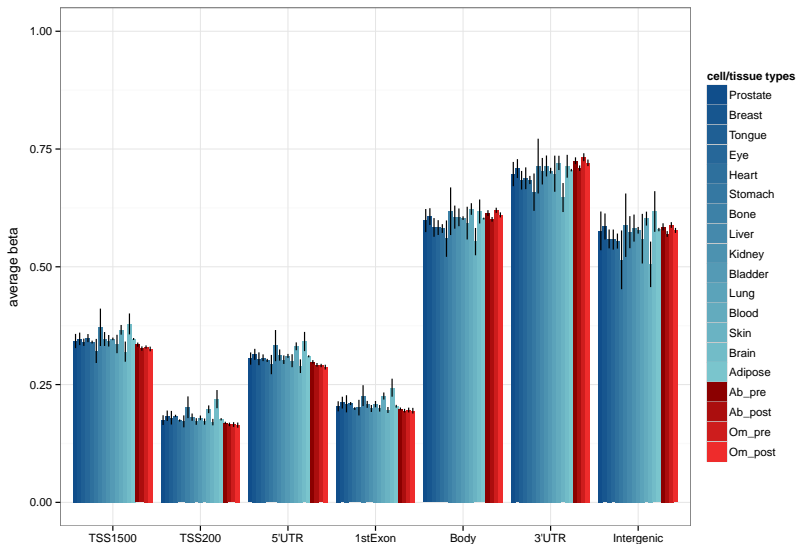


Investigating Methylation Profiles in Blood Cells

Publicly available data set (6 'healthy' males) - cell sorted



'Global' Methylation Profiles (MARMAL-AID)



mtDNA and Ion Torrent

Mitochondrial Sequencing



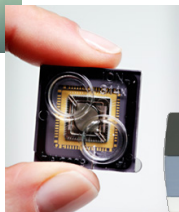
mtDNA: human ~16500bp

Ion Torrent PGM

Long range PCR: 2 amplicons

AUD\$400 per sample

**AUD\$80 per sample
(custom barcodes)**



Ion 316 chip: we can currently multiplex 48 complete mt genomes

Avg fold depth: 1500-2000

Acknowledgements

ESR/Wakefield Obesity Project: Dr Donia Macartney-Coxson, Prof Richard Stubbs, Angela Jones, Daniel Kay

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