Australian Genomics Health Alliance

# Program One Workshop: RNA-sequencing

6th September 2019

| Time | Topic | Presenter |
|---|---|---|
| 1:30 - 1:35 | Welcome and introduction | **Nigel Laing –** Centre for Medical Research, University of Western Australia and Harry Perkins Institute of Medical Research<br>**Hamish Scott** – Genetic and Molecular Pathology, Centre for Cancer Biology |
| **SESSION 1:** Chair - Nigel Laing | | |
| 1:35 – 1:50 | Diagnostic RNA-seq pipelines. | **Karin Kassahn** – Technology Advancement Unit, SA Pathology |
| 1:50 – 2:05 | RNA studies extend diagnostic yield by 20% in a paediatric neuromuscular cohort of 220 families. | **Sandra Cooper** – Kids Neuroscience Centre, University of Sydney |
| 2:05 – 2:25 | Using RNA-seq to determine treatment, monitor disease and classify risk in childhood leukaemia. | **Alicia Oshlack** – Bioinformatics, Murdoch Children's Research Institute |
| 2:25 – 2:45 | Incorporation of RNA-seq with whole exome sequencing and copy number variation reveals a higher yield of clinically relevant variants than whole exome sequencing alone. | **Susan Branford** – Genetic and Molecular Pathology, Centre for Cancer Biology |
| 2:45 – 3:05 | Transcriptome sequencing-based genetic testing of cardiomyocytes derived from patients with hypertrophic cardiomyopathy. | **Richard Bagnall** - Bioinformatics and Molecular Genetics, Centenary Institute |
| **3:05 – 3:20 Afternoon Tea** | | |
| **SESSION 2:** Chair - Hamish Scott | | |
| 3:20 - 3:35 | RNA-sequencing: understanding the data. | **Fathin Faiz** – Neurogenetics, Diagnostic Genomics, PathWest |
| 3:35 - 3:55 | RNA-seq data analysis in the context of ALL. | **Jacqueline Rehn** – South Australian Health and Medical Research Institute |
| 3:55 - 4:10 | Machine learning and cloud computing is empowering RNA-seq analysis. | **Natalie Twine** – Cloud-Computing Bioinformatics, CSIRO |
| 4:10 - 4:25 | Implementation in clinical laboratories. | **Natalie Thorne** – Bioinformatics, Melbourne Genomics Health Alliance |
| 4:25 - 5:00 | Discussion & action plans. | **Nigel Laing**<br>**Hamish Scott** |
| **5:00 Workshop close** | | |