Melbourne Genomics Health Alliance

Global knowledge. Individual care.

Variant Curation Workshop

Singapore, 29-30 Sept 2016

Proudly supported by **Centre for Personalised and Precision Health**

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Variant Curation Workshop, Singapore: 29 – 30 September 2016

Objectives

By the end of this workshop, participants will:

- Be aware of the utility of genomic testing in clinical practice;
- Understand when single gene, gene panel or genomic testing are indicated;
- Appreciate the complexity of variant classification;
- Be familiar with the curation process, from the bioinformatics pipeline through to clinical application.
- Be able to apply variant filtering processes
- Appreciate the importance of phenotyping and be able to curate and manage gene lists
- Be able to utilise population variant databases and in-silico software to assess functional effects and potential pathogenicity of variants
- Be able to utilise software and databases, such as LOVD and IGV in variant curation
- Be able to curate variants and apply findings in a clinical setting
- Be aware of ethical implications and need for informed consent in genomic testing

Participants

Approximately 20 participants anticipated. Professional areas to include:

- Geneticists
- Non-genetic clinical specialists
- Clinical scientists

Local contacts

- Roger Foo (mdcrfsy@nus.edu.sg)
- Denise Goh (denise_li_meng_goh@nuhs.edu.sg)
- Zenia Tiang (ztiang@gis.a-star.edu.sg)

Presenters

- Tiong Yang Tan (Clinical Geneticist, VCGS)
- Natalie Thorne (Bioinformatician, Melbourne Genomics)
- Sebastian Lunke (Head of Translational Genomics Unit, VCGS)
- Roger Foo, Denise Goh (NUH), and Saumya Jamuar (KKH)

Program

Day 1: Thursday 29 September: Variant Curation

Venue: MD6-10-01T [Lecture]; MD6-08-01U & MD6-08-02U [Practical]

Time	Topic	Duration (mins)	Speaker
9.00	Registration & coffee		
	The Fundamentals		
9.30	Welcome Introduction Singapore Genomics – local landscape	15	Roger Foo
9.45	Melbourne Genomics – Australian landscape	10	Natalie Thorne
9.55	Bioinformatics analysis	40	Natalie Thorne
10.35	*Morning tea*	*25*	
11.00	Curation overview	20	Tiong Tan
11.20	In-silico scores and conservation	30	Natalie Thorne
11.50	Variant classification LOVD database	45	Sebastian Lunke
12.35	*Lunch*	*45*	
	Practical elements of variant curation – Session 1*		
1.15	A. Searching population and locus specific databases	45	Natalie Thorne
	B. In-silico prediction scores	45	Sebastian Lunke
2.45	*Afternoon tea*	*15*	
	Practical elements of variant curation – Session 2*		
3.00	C. Variant filtering (including gene list curation and management)	45	Natalie Thorne
	D. Assessing functional effects of variants	45	Tiong Tan
	Instructive Case Study		
4.30	Case study – worked example of variant curation	30	Tiong Tan, Natalie Thorne, Sebastian Lunke
5.00	**CLOSE**		

* Two multidisciplinary groups to run concurrently in each session, with rotation into other group after 45 minutes.

Day 2: Friday 30 September: The Clinical-Laboratory Interface

Venue: MD6-10-01T [Lecture]; MD6-02-01E & MD6-02-01F [Practical]

Time	Topic	Duration (mins)	Speaker
	Exome sequencing in the Clinic		
9.30	Introduction	15	Tiong Tan
9.45	The importance of phenotyping – impact on test selection and gene lists	20	Tiong Tan
10.05	Impact on diagnosis and management, importance of pre-test genomic counselling	20	Tiong Tan
10.25	Exome/Genome reporting ▪ What to include ▪ What to not include ▪ Incidental findings (inc. consent and ethics)	30	Sebastian Lunke
10.55	*Morning tea*	*20*	
	Variant Curation in Action –practical sessions**		
11.15	Session 1 – 2 simplex cases	90	Roger Foo, Denise Goh, Saumya Jamuar Leader: Tiong Tan
12.45	*Lunch*	*40*	
1.25	Session 2 – 3 complex cases	90	Roger Foo, Denise Goh, Saumya Jamuar Leader: Sebastian Lunke
2.55	*Afternoon tea*	*15*	
	Genomics Q & A		
3.10	With questions from participants and option to present variants for discussion	60	Tiong Tan, Natalie Thorne, Sebastian Lunke, Roger Foo
4.10	Closing remarks	5	Roger Foo Tiong Tan
4.15	**CLOSE**		

**Variant Curation in Action:

- Session 1 – participants to work in groups of 4 to 6 (3 or 4 groups total) – each to curate two singleton cases from Phase 2 of Melbourne Genomics.
- Session 2 – participants to works in groups of 2 or 3 – each to curate 3 more complex cases, possibly involving trios, from Phase 2 of Melbourne Genomics.

Each session to be structured as:
5 minutes – session leader to present clinical cases, backgrounding phenotype and results of bioinformatics pipeline
55 minutes – groups to curate variants using LOVD+, searching population data bases, in-silico and functional analyses
30 minutes – simulated MDT (multi-disciplinary team) meeting – groups report on variant classification, with discussion relating to phenotype, clinical implications and counselling issues.