



Variant Effect Prediction Training Course

27- 30 August 2018

NUMed, Malaysia

PROGRAM

@ 8th August 2018

Monday 27th August

- 10:00 - 11:00 **REGISTRATION**
- 11:00 - 12:45 **PLENARY SESSION 1**
Iskandar Demonstration Theatre
- 11:00 - 11:15 **Welcome & Introduction**
- Johan den Dunnen
Leiden Univ. Medical Center, Leiden, Netherlands
&
Chris Baldwin
Deputy CEO, Dean of Biomedical and Foundation Sciences, Newcastle University Medicine
Malaysia
- 11:15 - 12:00 **Variants in the genome, position and possible consequences**
- Johan den Dunnen
Leiden Univ. Medical Center, Leiden, Netherlands
- 12:00 - 12:45 **Calling DNA variants using different tools**
- Raman Sethi
National University of Singapore, Singapore

12:45 - 14:00 **Lunch Break - Fame Cafe**

- 14:00 - 16:00 **PLENARY SESSION 2**
Iskandar Demonstration Theatre
- 14:00 - 14:45 **General databases: OMIM, dbSNP, gnomAD (ExAC), etc.**
- Robert Kuhn
UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA
- 14:45 - 16:00 **HGVS Nomenclature Workshop**
- Johan den Dunnen
Leiden Univ. Medical Center, Leiden, Netherlands

16:00 - 16:30 *Coffee Break - Fame Cafe*

- 16:30 - 18:00 **PLENARY SESSION 3**
Iskandar Demonstration Theatre
- Human Phenotype Ontology (HPO) Workshop**
- Andreas Laner
MGZ - Medical Genetics Centre, Munich, Germany
&
Johan den Dunnen
Leiden Univ. Medical Center, Leiden, Netherlands

18:00 **END OF DAY**

Tuesday 28th August

- 8:30 - 10:30 **PLENARY SESSION 4**
Iskandar Demonstration Theatre
- 8:30 - 9:00 **Gene Variant Databases (LSDBs): HGMD, LOVD, ClinVar, etc.**
- Johan den Dunnen
Leiden Univ. Medical Center, Leiden, Netherlands
- 9:00 - 9:45 **Variant Classification: ACMG recommendations**
- Andreas Laner & Anna Benet-Pages
MGZ - Medical Genetics Centre, Munich, Germany
- 9:45 - 10:30 **Viewing the Data: the Ensembl browser and its possibilities**
- Ben Moore
Ensembl, EMBL - EBI, Cambridge, UK

10:30 - 11:00 *Coffee Break - Fame Cafe*

11:00 - 11:30

PLENARY SESSION 5
Iskandar Demonstration Theatre

Samples to answer: Developing a cost-effective and robust targeted NGS workflow

LEE Chee Yang
Senior Field Application Scientist - Genomics Solution Division, Diagnostics & Genomics Group, South Asia Pacific, Agilent Technologies Singapore
Agilent Technologies Singapore is a sponsor of the 3rd Variant Effect Prediction Training Course

11:30 - 13:00

WORKSHOP STREAM A
IT Cluster 1

Variant Interpretation
Using ACMG Guidelines

Andreas Laner & Anna Benet-Pages

WORKSHOP STREAM B
IT Cluster 2

Ensembl Genome Browser
Workshop

Ben Moore

13:00 - 14:15

Lunch Break - Fame Cafe

14:15 - 15:45

WORKSHOP STREAM A
IT Cluster 2

Ensembl Genome Browser
Workshop

Ben Moore

WORKSHOP STREAM B
IT Cluster 1

LOVD+
Demonstration

Johan den Dunnen

15:45 - 16:30

Coffee Break & Poster Session 1 - Fame Cafe

16:30 - 18:00

WORKSHOP STREAM A
IT Cluster 1

LOVD+
Demonstration

Johan den Dunnen

WORKSHOP STREAM B
IT Cluster 2

Variant Interpretation
Using ACMG Guidelines

Andreas Laner & Anna Benet-Pages

18:00

END OF DAY

Wednesday 29th August

8:30 - 10:30

PLENARY SESSION 6
Iskandar Demonstration Theatre

8:30 - 9:15

Viewing the Data: the UCSC browser and its possibilities

Robert Kuhn
UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA

9:15 - 10:00	Variant Annotation: VEP	
	Ben Moore Ensembl, EMBL - EBI, Cambridge, UK	
10:00 - 10:30	Copy-number variation detection from exon capture data	
	Anna Benet-Pages MGZ - Medical Genetics Centre, Munich, Germany	
10:30 - 10:45	Assemble for Group Photograph Go to "Newcastle University Medicine Malaysia" Arch at Bells Court	
10:45 - 11:15	Coffee Break - Fame Cafe	
11:15 - 12:45	PLENARY SESSION 7 Iskandar Demonstration Theatre	
11:15 - 12:00	Potential Consequences on the RNA Level and using prediction tools	
	Andreas Laner MGZ - Medical Genetics Centre, Munich, Germany	
12:00 - 12:45	Potential Consequences on Protein Level and using prediction tools	
	Poh San Lai National University of Singapore	
12:45 - 14:00	Lunch Break - Fame Cafe	
14:00 - 15:30	WORKSHOP STREAM A IT Cluster 1	WORKSHOP STREAM B IT Cluster 2
	UCSC Genome Browser	Variant Annotation using VEP
	Robert Kuhn	Ben Moore
15:30 - 16:15	Coffee Break & Poster Session 2 - Fame Cafe	
16:15 - 17:45	WORKSHOP STREAM A IT Cluster 2	WORKSHOP STREAM B IT Cluster 1
	Variant Annotation using VEP	UCSC Genome Browser
	Ben Moore	Robert Kuhn
17:45	END OF DAY	

Thursday 30th August

8:30 - 10:30	PLENARY SESSION 7 Iskandar Demonstration Theatre
8:30 - 9:15	Options for functional testing of candidate variants
	Bruno Reversade Institute of Medical Biology, Singapore

9:15 - 10:00	UCSC Variant Annotation Integrator Robert Kuhn UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA
10:00 - 10:30	NGS in diagnostics: where things can go wrong Anna Benet-Pages MGZ - Medical Genetics Centre, Munich, Germany & Johan den Dunnen Leiden Univ. Medical Center, Leiden, Netherlands
10:30 - 11:00	Coffee Break - Fame Cafe
11:00 - 13:00	PLENARY SESSION 8 Iskandar Demonstration Theatre
11:00 - 11:45	Setting up diagnostic NGS application in Malaysia Zilfalil bin Alwi Universiti Sains Malaysia, Kelantan, Malaysia
11:45 - 12:15	Future developments Johan den Dunnen Leiden Univ. Medical Center, Leiden, Netherlands
12:15 - 13:00	Q&A, Course Evaluation and Close
13:00	COURSE END