



Variant Effect Prediction Training Course

27- 30 August 2018

NUMed, Malaysia

PROGRAM

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 UCSC Genome Browser Robert Kuhn	WORKSHOP STREAM B IT Cluster 2 Variant Annotation using VEP Ben Moore
15:30 - 16:15	Coffee Break & Poster Session 2 - Fame Cafe	
16:15 - 17:45	WORKSHOP STREAM A IT Cluster 2 Variant Annotation using VEP Ben Moore	WORKSHOP STREAM B IT Cluster 1 UCSC Genome Browser Robert Kuhn
17:45	END OF DAY	

Thursday 30th August

- 8:30 - 10:30 **PLENARY SESSION 7**
Iskandar Demonstration Theatre
- 8:30 - 8:50 **Functional Testing from lab tests to animal models**

Johan den Dunnen
Leiden Univ. Medical Center, Leiden, Netherlands
- 8:50 - 9:35 **UCSC Variant Annotation Integrator**

Robert Kuhn
UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA
- 9:35 - 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**