



Variant Effect Prediction Training Course

27- 30 April 2018

NUMed, Malaysia

PROGRAM

DRAFT 27 APRIL 2018

TBC = To be Confirmed

TBA = To be Announced

Program subject to changes, additions, subtractions and rearrangement

Monday 27th August

9:30 - 11:00 **REGISTRATION**

11:00 - 12:45 **PLENARY SESSION 1**
Iskandar Demonstration Theatre

11:00 - 11:15 **Welcome & Introduction**

Johan T. den Dunnen
Leiden Univ. Medical Center, Leiden, Netherlands

11:15 - 12:00 **Variants in the genome, position and possible consequences**

TBA

12:00 - 12:45 **Limitations from sequencing technologies: Calling DNA variants - SNV, CNV & SV**

Raman Sethi (TBC)
National University of Singapore, Singapore

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

14:00 - 16:00	PLENARY SESSION 2 Iskandar Demonstration Theatre		
14:00 - 14:45	Limitations from sequencing technologies: general databases, OMIM, dbSNP, ExAC etc. Robert Kuhn UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA		
14:45 - 15:30	Human Phenotype Ontology (HPO) Swati Tomar (TBC) National University of Singapore, Singapore		
15:30 - 16:00	DNA Variants, Phenotypes and Databases: HGMD, LOVD, ClinVar etc & the importance of sharing information Johan den Dunnen Leiden Univ. Medical Center, Leiden, Netherlands		
16:00 - 16:30	Coffee Break		
16:30 - 18:00	CONCURRENT PRACTICAL Iskandar Demonstration Theatre HGVS, HPO and submitting Data Workshop Johan T. den Dunnen	CONCURRENT PRACTICAL Seminar Room 2.2 TBA	CONCURRENT PRACTICAL Seminar Room 2.3 TBA
18:00	END OF DAY		

Tuesday 28th August

8:30 - 10:45	PLENARY SESSION 3 Iskandar Demonstration Theatre
8:30 - 9:15	HGVS Nomenclature - describing variants Johan den Dunnen Leiden Univ. Medical Center, Leiden, Netherlands
9:15 - 10:00	Variant Classification: ACMG recommendations Andreas Laner & Anna Benet-Pages MGZ - Medical Genetics Centre, Munich, Germany
10:00 - 10:30	Viewing the Data: the Ensembl browser and its possibilities Speaker TBA Ensembl, EMBL - EBI, Cambridge, UK

10:30 - 10:45	Company Presentation Agilent Title TBA		
10:45 - 11:30	Exhibits Open & Coffee Break		
11:30 - 13:00	CONCURRENT PRACTICAL Iskandar Demonstration Theatre Variant Interpretation Using ACMG Guidelines Andreas Laner & Anna Benet-Pages	CONCURRENT PRACTICAL Seminar Room 2.2 Ensembl Genome Browser Workshop (beginners) Speaker TBA	
13:00 - 14:15	Lunch Break		
14:15 - 15:45	CONCURRENT PRACTICAL Iskandar Demonstration Theatre Variant Interpretation Using ACMG Guidelines (Repeated) Andreas Laner & Anna Benet-Pages	CONCURRENT PRACTICAL Seminar Room 2.2 Ensembl Genome Browser Workshop (advanced) Speaker TBA	
15:45 - 16:30	Exhibits Open & Poster Session 1		
16:30 - 18:00	CONCURRENT PRACTICAL Iskandar Demonstration Theatre HGVS, HPO and submitting Data Workshop (Repeated) Johan T. den Dunnen	CONCURRENT PRACTICAL Seminar Room 2.2 TBA	CONCURRENT PRACTICAL Seminar Room 2.3 TBA
18:00	END OF DAY		

Wednesday 29th August

8:30 - 10:30	PLENARY SESSION 4 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
	Speaker TBA 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 - 11:15 Exhibits Open & Poster Session 2

11:15 - 12:45 **PLENARY SESSION 5** **Iskandar Demonstration Theatre**

11:15 - 12:00 **Potential Consequences on the RNA Level**

Andreas Laner
MGZ - Medical Genetics Centre, Munich, Germany

12:00 - 12:45 **Potential Consequences on Protein Level & tools that bridge the evaluation of the consequences of variants between DNA and protein**

Poh San Lai
National University of Singapore

12:45 - 14:00 Lunch Break

14:00 - 15:30	CONCURRENT PRACTICAL Iskandar Demonstration Theatre	CONCURRENT PRACTICAL Seminar Room 2.2	CONCURRENT PRACTICAL Seminar Room 2.3
	UCSC Genome Browser (beginner)	TBA	TBA
	Robert Kühn	TBA	TBA

15:30 - 16:15 Exhibits Open & Coffee Break

16:15 - 17:45	CONCURRENT PRACTICAL Iskandar Demonstration Theatre	CONCURRENT PRACTICAL Seminar Room 2.2	CONCURRENT PRACTICAL Seminar Room 2.3
	UCSC Genome Browser (advanced)	TBA	TBA
	Robert Kühn	TBA	TBA

17:45 **END OF DAY**

Thursday 30th August

8:30 - 10:30	PLENARY SESSION 6 Iskandar Demonstration Theatre
8:30 - 9:15	Functional Testing: lab tests, animal models ... Bruno Reversade (TBC) Institute of Medical Biology, Singapore
9:15 - 10:00	TBA
10:00 - 10:30	TBA
10:30 - 11:00	Coffee Break
11:00 - 13:00	PLENARY SESSION 7 Iskandar Demonstration Theatre
11:00 - 11:45	Prediction Tools RNA & Protein Swati Tomar (TBC) National University of Singapore, Singapore
11:45 - 12:30	Diagnostic NGS application in Malaysia Zilfalil bin Alwi Universiti Sains Malaysia, Kelantan, Malaysia
12:30 - 13:00	Future Developments, Course Evaluation and Close
13:00	COURSE END