Variant Effect Prediction Training Course

6 - 8 November 2017

Prague, Czech Republic

DRAFT PROGRAM (23 August 2017)

(subject to changes)
All session times to be confirmed
TBA = To be Announced1

Monday 6th November	
7.30- 8.30	**REGISTRATION**
8.30 - 10.15	**PLENARY SESSION 1** **Room: Zürich 1-2**
8.30 - 8.45	**Welcome & Introduction** Johan T. den Dunnen *Leiden Univ. Medical Center, Leiden, Netherlands*
8.45 - 9.30	**Variants in the genome, position and possible consequences** Jan Traeger-Synodinos *Dept. of Medical Genetics, National and Kapodistrian University of Athens, Greece*
9.30 - 10.15	**Calling DNA variants - SNV, CNV & SV** Steven Laurie *RD Connect*
10.15 - 11.00	**Coffee Break & Poster Session**
11.00 - 12.30	**PLENARY SESSION 2** **Room: Zürich 1-2**
11.00 - 11.45	**Human Phenotype Ontology (HPO)** Sebastian Köhler *NeuroCure Cluster of Excellence, Charité Universitätsklinikum Berlin, Germany*
11.45 - 12.30	**Gene variant databases & sharing information** Martina Witsch Baumgartner *Dept.für Medizinische Genetik, Molekulare und Klinische Pharmakologie* *Medizinische Universität Innsbruck, Austria*

12.30 -13.30	**Lunch**		
13.30 - 15.15	**PLENARY SESSION 3** **Room: Zürich 1-2**		
13.30 - 14.00	**HGVS Nomenclature - describing variants** Johan den Dunnen *Leiden Univ. Medical Center, Leiden, Netherlands*		
14.00 - 14.30	**Variant Classification: ACMG recommendations** Andreas Laner *MGZ - Medical Genetics Centre, Munich, Germany*		
14.30 - 15.15	**Viewing the Data: the Ensembl browser and its possibilities** Benjamin Moore *Ensembl, EMBL - EBI, Cambridge, UK*		
15.15 - 15.45	**Coffee Break**		
15.45 - 17.15	**CONCURRENT PRACTICAL** **Room: Zürich 1-2** **Ensembl Genome Browser** Benjamin Moore	**CONCURRENT PRACTICAL** **Room: Zürich 3** **ACMG classification** Andreas Laner	**CONCURRENT PRACTICAL** **Room: Zürich 4** **Sponsored Practical: Sophia Genetics** Speaker TBA
17.15 - 17.30	**Refresh Break**		
17.30 - 19.00	**CONCURRENT PRACTICAL** **Room: Zürich 1-2** **Ensembl Genome Browser** Benjamin Moore	**CONCURRENT PRACTICAL** **Room: Zürich 3** **Sponsored Practical: Qiagen** Speaker TBA	**CONCURRENT PRACTICAL** **Room: Zürich 4** **HGVS, HPO and submitting Data Workshop (once only, *this is not repeated*)** *(*see description on last page)* Johan T. den Dunnen
19.15 - 21.00	**WELCOME RECEPTION - NH PRAGUE CITY**		

Tuesday 7th November

8.30 - 10.30	**PLENARY SESSION 4** **Room: Zürich 1-2**
8.30 - 9.15	**Viewing the Data: the UCSC browser and its possibilities** Robert Kühn *UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA*
9.15 - 10.00	**Variant Annotation: VEP** Benjamin Moore *Ensembl, EMBL - EBI, Cambridge, UK*
10.00 - 10.30	**The RD-Connect platform** Steve Laurie *RD Connect*
10.30 - 11.15	**Coffee Break**
11.15- 13.00	**PLENARY SESSION 5** **Room: Zürich 1-2**
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** Jana Marie Schwarz / Sebastian Köhler *NeuroCure Cluster of Excellence, Charité Universitätsklinikum Berlin, Germany*
12.45 - 13.15	**Copy-Number Variation Detection From Exon Capture Data** Ana Benet Pages *MGZ - Medical Genetics Centre, Munich, Germany*
13.15 - 14.15	**Lunch Break**

14.15 - 15.45	**CONCURRENT PRACTICAL** **Room: Zürich 1-2**	**CONCURRENT PRACTICAL** **Room: Zürich 3**	**CONCURRENT PRACTICAL** **Room: Zürich 4**
	Sponsored Practical: Sophia Genetics Speaker TBA	**Using the RD-Connect Genomics platform to solve rare disease cases** Steven Laurie	**UCSC Genome Browser** Robert Kühn

15.45 - 16.15	**Coffee Break**

16.15 - 17.45	**CONCURRENT PRACTICAL** **Room: Zürich 1-2** **ACMG classification** Andreas Laner	**CONCURRENT PRACTICAL** **Room: Zürich 3** **Using the RD-Connect Genomics platform to solve rare disease cases** Steven Laurie	**CONCURRENT PRACTICAL** **Room: Zürich 4** **Sponsored Practical: Limbus Medical Technology (once only, *this is not repeated*)** Speaker TBA
19.00 -	**CONFERENCE DINNER TO BE ADVISED**		

Wednesday 8th November

8.30 - 10.30 **PLENARY SESSION 5**
Room: Zürich 1-2

8.30- 9.15 **Prioritise, Annotate and Filter Variants**

Christophe Béroud
RD Connect

9.15 - 10.00 **Complex penetrance analysis on a model of CFTR locus specific efforts**

Milan Macek Jr.
Charles University, Prague, Czech Republic

10.00 - 10.30 **TBA**

10.30 - 11.00 **Coffee Break**

11.00 - 13.15 **PLENARY SESSION 6**
Room: Zürich 1-2

11.00 - 11.45 **Functional Testing: lab tests, animal models …**

Speaker TBA

11.45 - 13.15

CONCURRENT PRACTICAL Room: Zürich 1-2	CONCURRENT PRACTICAL Room: Zürich 3	CONCURRENT PRACTICAL Room: Zürich 4
Sponsored Practical: Qiagen	**MutationTaster & RegulationSpotter**	**VarAFT & UMD Predictor (RD connect)**
Speaker TBA	Jana Marie Schwartz & Sebastian Kohler	Christophe Béroud

13.15- 14.15 **Lunch Break**

14.15 - 15.45

CONCURRENT PRACTICAL Room: Zürich 1-2	CONCURRENT PRACTICAL Room: Zürich 3	CONCURRENT PRACTICAL Room: Zürich 4
UCSC Genome Browser	**MutationTaster & RegulationSpotter**	**VarAFT & UMD Predictor (RD connect)**
Robert Kühn	Jana Marie Schwartz & Sebastian Kohler	Christophe Béroud

15.45 - 16.15 **Coffee Break**

16.15 - 18.00 **PLENARY SESSION 7**
Room: Zürich 1-2

16.15 - 16.45 **The Role of Rare Variants in Complex Phenotypes**

Stanislav Kmoch
Charles University in Prague and General University Hospital in Prague, Czech Republic

16.45- 17.30 **NGS in Diagnostics: a practical example in hereditary cardiomyopathies**

Patricia Norambuena
University Hospital Motol , Prague, Czech Republic

17.30 - 18.00 **Future Developments, Meeting Evaluation and Close**

Speaker TBA

18.00 COURSE END

***HGVS, HPO and submitting Data Workshop**

DNA diagnostics is based on sharing data on genes, variants and phenotypes. Without sharing DNA diagnostics would not be possible. When we do not share, we do not offer optimal care to the patients and their families. In this practical we will demonstrate how you can share your data by performing an actual database submission. Phenotype will be described using the Human Phenotype Ontology (HPO) and sequence variant using the HGVS recommendations. After submission, based on the experience gained, we will query the database to determine whether a specific variant is disease-associated or not.

SCHEDULE OF PRACTICAL DEMONSTRATIONS (session time still subject to change)

PRESENTATION	MONDAY 15.45 - 17.15	MONDAY 17.30 - 19.00	TUESDAY 14.15 - 15.45	TUESDAY 16.15 - 17.45	WEDNESDAY 11.45 - 13.15	WEDNESDAY 14.15 - 15.45
Ensemble Genome Browser	X	X				
UCSC Genome Browser			X			X
ACMG Classification	X			X		
MutationTaster & Regulation Spotter					X	X
VarAFT & UMD Predictor					X	X
RD Connect Platform			X	X		
*HGVS, HPO & submitting Data		X				
Qiagen		X			X	
Sophia Genetics			X	X		
Limbus Med. Tech.	X					