Variant Effect Prediction Training Course

6 - 8 November 2017

Prague, Czech Republic

PROGRAM (3 Oct. 2017)

(subject to minor changes)
TBA = To be Announced

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**	**CONCURRENT PRACTICAL** **Room: Zürich 3**	**CONCURRENT PRACTICAL** **Room: Zürich 4**
	Ensembl Genome Browser Benjamin Moore	**ACMG classification** Andreas Laner	**Sponsored Practical: Sophia Genetics** Gaetano Bonifacio & Jean-François Vanbellinghen

17.15 - 17.30 **Refresh Break**

17.30 - 19.00	**CONCURRENT PRACTICAL** **Room: Zürich 1-2**	**CONCURRENT PRACTICAL** **Room: Zürich 3**	**CONCURRENT PRACTICAL** **Room: Zürich 4**
	Ensembl Genome Browser Benjamin Moore	**Sponsored Practical: Qiagen** Ruth Burton	**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 Kuhn *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, CNAG - CRG, Barcelona, Spain*		
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 *NeuroCure Cluster of Excellence, Charité Universitätsklinikum Berlin, Germany*		
12.45 - 13.15	**CNV detection from targeted next-generation panel sequencing data in routine diagnostics** Anna Benet Pages *MGZ - Medical Genetics Centre, Munich, Germany*		
13.15 - 14.15	**Lunch Break**		
14.15 - 15.45	**CONCURRENT PRACTICAL** **Room: Zürich 1-2** **Sponsored Practical: Sophia Genetics** Gaetano Bonifacio & Jean-François Vanbellinghen	**CONCURRENT PRACTICAL** **Room: Zürich 3** **Using the RD-Connect Genomics platform to solve rare disease cases** Steven Laurie	**CONCURRENT PRACTICAL** **Room: Zürich 4** **UCSC Genome Browser** Robert Kuhn
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 Technologies (once only, *this is not repeated*)** **Clinical diagnostics with automated SNP and CNV analysis using VARVIS** Konrad Platzer, Institute of Human Genetics, Univ. Hospital of Leipzig Yvonne Schmitz, Limbus Medical Technologies

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, Aix-Marseille, Univ. Marseille, France

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)**
	Ruth Burton	Jana Marie Schwartz & Daniela Hombach	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 Kuhn	Jana Marie Schwartz & Daniela Hombach	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**

Anna Benet Pages & Andreas Laner
MGZ - Medical Genetics Centre, Munich, Germany

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		