



POCKET PROGRAM



THE
HUMAN VARIOME
PROJECT

an NGO official partner of UNESCO
sharing data · reducing disease

6 – 8 Nov 2017
Prague
Czech Republic

2nd Variant Effect
Prediction Training
Course



The Human Variome Project and the Scientific Organising Committee wishes to express its gratitude to the following sponsors for their generous support of this event.

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IN KIND



NATIONAL CENTER
FOR
MEDICAL GENOMICS



RDConnect



2nd Variant Effect Prediction Training Course

6 – 8 November 2017

**NH Prague City Hotel
Prague
Czech Republic**



Scientific Programme Committee

Prof. Johan T. den Dunnen (Leiden, Nederland) CHAIR
Prof. Milan Macek Jr. (Prague, Czech Rep.) LOCAL ORGANISER
Dr Stanislav Knoch (Prague, Czech Rep.)
Dr Sebastian Kohler (Berlin, Germany)
Dr Andreas Laner (Munich, Germany)
Dr Joanne Traeger-Synodinos (Athens, Greece)



Monday 6th November

7.30- 8.30

REGISTRATION

Zürich Foyer

8.30 - 10.15

PLENARY SESSION 1

Room: Zürich I-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, CNAG-CRG, Barcelona, Spain

10.15 - 11.00

Coffee Break & Poster Session

11.00 - 12.30

PLENARY SESSION 2

Room: Zürich I-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





Monday 6th November

12.30 - 13.30 Lunch

13.30 - 15.15 **PLENARY SESSION 3**
Room: Zürich 1-2

13.30 - 14.00 **The HGVS recommendations to describe sequence variants**
Johan T. 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	Variant interpretation using ACMG guidelines	Sponsored Practical: Sophia Genetics
	Benjamin Moore	Andreas Laner	Gaetano Bonifacio & Jean-François Vanbellinghen

17.15 - 17.30 Refresh Break



Monday 6th November

17.30 - 19.00

CONCURRENT PRACTICAL

Room: Zürich 1-2

**Ensembl Genome
Browser**

Benjamin Moore

CONCURRENT PRACTICAL

Room: Zürich 3

**Sponsored
Practical:
Qiagen**

Ruth Burton
& Kamil Matulka

CONCURRENT PRACTICAL

Room: Zürich 4

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

Johan T. den Dunnen



19.15 - 21.00

WELCOME RECEPTION - NH PRAGUE CITY

**(once only, this is not repeated)*



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 - 13.15

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

CNV detection from targeted next-generation panel sequencing data in routine diagnostics

Anna Benet Pages

MGZ - Medical Genetics Centre, Munich, Germany



Tuesday 7th November

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

**Variant
interpretation
using ACMG
guidelines**

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:
Clinical diagnostics
with automated
SNP and CNV
analysis using
VARVIS**

Konrad Platzer &
Yvonne Schmitz

***(once only, this is not repeated)**

Wednesday 8th November

8.45 - 10.30

PLENARY SESSION 6

Room: Zürich I-2

8.45 - 9.30

Variants prioritization: annotation and filtration steps

Christophe Bérout

RD Connect, Aix-Marseille Univ., Marseille, France

9.30 - 10.15

Complex penetrance analysis on a model of CFTR locus specific efforts

Milan Macek Jr.

Charles University, Prague, Czech Republic

10.15 - 10.30

Assemble for group photograph

10.30 - 11.00

Coffee Break



11.00 - 13.15

PLENARY SESSION 7

Room: Zürich I-2

11.00 - 11.45

Functional Testing: from lab tests to animal models

Johan T. den Dunnen

Leiden Univ. Medical Center, Leiden, Netherlands

11.45 - 13.15

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Room: Zürich I-2

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Practical:
Qiagen**

Ruth Burton
& Kamil Matulka

CONCURRENT PRACTICAL

Room: Zürich 3

**MutationTaster
& Regulation
Spotter**

Jana Marie Schwarz
& Daniela Hombach

CONCURRENT PRACTICAL

Room: Zürich 4

**VarAFT & UMD
Predictor
(RD connect)**

Christophe Bérout

Wednesday 8th November

13.15- 14.15 Lunch Break

14.15 - 15.45

**CONCURRENT
PRACTICAL**

Room: Zürich 1-2

**UCSC Genome
Browser**

Robert Kuhn

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Christophe Bérout



15.45 - 16.15

Coffee Break

16.15 - 17.30

PLENARY SESSION 8

Room: Zürich 1-2

16.15 - 16.45

NGS in Diagnostics: a practical example in hereditary cardiomyopathies

Patricia Norambuena

University Hospital Motol, Prague, Czech Republic

16.45- 17.30

Future Developments, Meeting Evaluation and Close

Anna Benet Pages & Andreas Laner

MGZ - Medical Genetics Centre, Munich, Germany

17.30

COURSE END

SCHEDULE OF PRACTICAL DEMONSTRATIONS

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		



Meeting
Makers

e: info@meeting-makers.com

w: www.meeting-makers.com

