



# Variant Effect Prediction Training Course

## 29 - 31 May 2019

### Moscow, Russian Fed.

**DRAFT PROGRAM (2 Nov. 2018)**

**TBA = To be Announced TBC = To be Confirmed**

**Subject to additions, subtractions and rearranging.**

**All the workshops will probably be rearranged to different times**

### Wednesday 29th May

<b>8:00 - 9:00</b>	<b>REGISTRATION</b>
<b>9:00 - 10:40</b>	<b>PLENARY SESSION 1</b> <b>Room:</b>
<b>9:00 - 9:10</b>	<b>Welcome &amp; Introduction</b>  Johan T. den Dunnen Leiden Univ. Medical Center, Leiden, Netherlands
<b>9:10 - 9:55</b>	<b>Variants in the genome, position and possible consequences</b>  TBA
<b>9:55 - 10:40</b>	<b>NGS limitations: sequencing technologies/ calling variants</b>  TBA

**10:40 - 11:10 Coffee Break**

**11:10 - 13:05 PLENARY SESSION 2**  
**Room:**

**11:10 - 11:55 General databases: OMIM, dbSNP, gnomAD (ExAC), etc.**

Robert Kuhn  
UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA

**11:55 - 12:35**    **Gene Variant databases (LSDBs): HGMD, LOVD, ClinVar etc.**  
TBA

**12:35 - 13:05**    **Sponsored Company Lecture**  
TBA

**13:05 - 14:10**    **Lunch**

**14:10 - 15:30**    **PLENARY SESSION 3**  
**Room:**

**14:10 - 14:50**    **Variant Classification: ACMG recommendations**  
Andreas Laner  
MGZ - Medical Genetics Centre, Munich, Germany

**14:50 - 15:30**    **Viewing the Data: the Ensembl browser and its possibilities**  
TBA  
Ensembl, EMBL - EBI, Cambridge, UK

**15:30 - 16:15**    **Coffee Break & Poster Session**

**16:15 - 17:30**    **PLENARY SESSION 4**  
**Room:**

**16:15 - 17:30**    **HGVS Nomenclature: describing variants - WORKSHOP**  
Johan den Dunnen  
Leiden Univ. Medical Center, Leiden, Netherlands

**17:30 - 17:45**    **Refresh Break**

**17:45 - 19:00**    **PLENARY SESSION 5**  
**Room:**

**17:45 - 19:00**    **NGS in Diagnostics: where things can go wrong - WORKSHOP**  
Anna Benet Pàges  
MGZ - Medical Genetics Centre, Munich, Germany  
&  
Johan den Dunnen  
Leiden Univ. Medical Center, Leiden, Netherlands

**19.15 - 21.00**    **WELCOME RECEPTION TBC**

## Thursday 30th May

### 8:30 - 10:30 **PLENARY SESSION 6**

**Room:**

#### 8:30 - 9:10 **Viewing the Data: the UCSC browser and its possibilities**

Robert Kuhn

UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA

#### 9:10 - 9:40 **Variant Annotation: VEP**

TBA

Ensembl, EMBL - EBI, Cambridge, UK

#### 9:40 - 10:10 **Sponsored Company Lecture**

TBA

#### 10:10 - 10:20 **Assemble for Group Photograph**

### 10:20 - 11:00 **Coffee Break**

### 11:00 - 13:00 **PLENARY SESSION 7**

**Room:**

#### 11:00 - 11:45 **Potential Consequences on the RNA Level and using prediction tools**

Andreas Laner

MGZ - Medical Genetics Centre, Munich, Germany

#### 11:45 - 12:30 **Potential Consequences on Protein Level and using prediction tools**

TBA

#### 12:30 - 13:00 **Copy Number Variations from Whole Exome Sequencing**

Anna Benet Pages

MGZ - Medical Genetics Centre, Munich, Germany

### 13:00 - 14:00 **Lunch Break**

#### 14:00 - 15:30 **WORKSHOP STREAM A**

**Room:**

##### **ACMG classification**

Andreas Laner & Anna  
Benet Pàges

#### 14:00 - 15:30 **WORKSHOP STREAM B**

**Room:**

##### **Ensembl Genome Browser**

TBA

#### 14:00 - 15:30 **WORKSHOP STREAM C**

**Room:**

##### **UCSC Genome Browser**

Robert Kuhn

### 15:30 - 16:00 **Coffee Break**

16:00 - 17:30	<b>WORKSHOP STREAM A</b> <b>Room:</b>  <b>Ensembl Genome Browser</b>  TBA	<b>WORKSHOP STREAM B</b> <b>Room:</b>  <b>UCSC Genome Browser</b>  Robert Kuhn	<b>WORKSHOP STREAM C</b> <b>Room:</b>  <b>ACMG classification</b>  Andreas Laner & Anna Benet Páges
17:30 - 17:45 Refresh Break			
17:45 - 19:15	<b>WORKSHOP STREAM A</b> <b>Room:</b>  <b>UCSC Genome Browser</b>  Robert Kuhn	<b>WORKSHOP STREAM B</b> <b>Room:</b>  <b>ACMG classification</b>  Andreas Laner & Anna Benet Páges	<b>WORKSHOP STREAM C</b> <b>Room:</b>  <b>Ensembl Genome Browser</b>  TBA
19:15	<b>END OF DAY</b>		

## Friday 31 May

8:30 - 10:30 **PLENARY SESSION 8**

**Room:**

8:30 - 9:00 **Functional Testing: Options for functional testing of candidate variants**

TBA

9:00 - 9:45 **Variant Annotation: Variant Annotation Integrator (VAI)**

Robert Kuhn

UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA

9:45 - 10:30 **NGS in practice: local Moscow lab**

TBA

10:30 - 11:00 **Coffee Break**

### CONCURRENT WORKSHOPS SET A

11:00 - 12:30 **CONCURRENT WORKSHOP 1**

**Room:**

**WES analysis using LOVD+**

Johan den Dunnen

**CONCURRENT WORKSHOP 2**

**Room:**

**VAI**

Robert Kuhn

**CONCURRENT WORKSHOP 3**

**Room:**

**Sponsored Company Workshop**

TBA

12:30 - 13:30 **Lunch Break**

### CONCURRENT WORKSHOPS SET B

13:30 - 15:00 **CONCURRENT WORKSHOP 4**

**Room:**

**VEP**

TBA (Ensembl)

**CONCURRENT WORKSHOP 5**

**Room:**

**Sponsored Company Workshop**

TBA

**CONCURRENT WORKSHOP 6**

**Room:**

**HGVS, HPO and submitting Data**

Johan den Dunnen

15:00 - 15:30 **Coffee Break**

## CONCURRENT WORKSHOPS SET C

<b>15:30 - 17:00</b>	<b>CONCURRENT WORKSHOP 7</b> <b>Room:</b>  <b>HGVS, HPO and submitting Data</b>  Johan den Dunnen	<b>CONCURRENT WORKSHOP 8</b> <b>Room:</b>  <b>VEP</b>  TBA (Ensembl)	<b>CONCURRENT WORKSHOP 9</b> <b>Room:</b>  <b>Sponsored Company Workshop</b>  TBA
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**17:00 - 17:10 Refresh Break**

**17:10 - 18:10 PLENARY SESSION 9**  
**Room:**

**17:10 - 17:40 Future Developments**

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

**17:40 - 18:15 Meeting Evaluation and Close**

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

**18:15 COURSE END**