



Variant Effect Prediction Training Course

29 - 31 May 2019
Moscow, Russian Fed.

DRAFT PROGRAM (29 Jan. 2019)

TBA = To be Announced TBC = To be Confirmed

Subject to additions, subtractions and rearranging.

**EVERYONE TO BE ALLOCATED A STREAM
PLEASE STAY IN YOUR STREAM FOR WORKSHOPS**

Wednesday 29th May

7:30 - 8:30	REGISTRATION
8:30 - 10:10	PLENARY SESSION 1 Room: Tretyakov+Morozov
8:30 - 8:40	Welcome & Introduction Johan T. den Dunnen Leiden Univ. Medical Center, Leiden, Netherlands
8:40 - 9:20	Variants in the genome, position and possible consequences TBA
9:20 - 10:10	NGS limitations: sequencing technologies/ calling variants TBA
10:10 - 10:40	Coffee Break
10:40 - 12:35	PLENARY SESSION 2 Room: Tretyakov+Morozov
10:40 - 11:20	General databases: OMIM, dbSNP, gnomAD (ExAC), etc. Robert Kuhn UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA

11:20 - 12:00 **Gene Variant databases (LSDBs): HGMD, LOVD, ClinVar etc.**

TBA

12:00 - 12:40 **Variant Classification: ACMG recommendations**

Andreas Laner
MGZ - Medical Genetics Centre, Munich, Germany

12:40 - 13:40 **Lunch**

13:40 - 15:40 **PLENARY SESSION 3**

Room: Tretyakov+Morozov

13:40 - 14:20 **Viewing the Data: the Ensembl browser and its possibilities**

Benjamin Moore
Ensembl, EMBL - EBI, Cambridge, UK

14:20 - 15:00 **Variant Annotation: VEP**

Benjamin Moore
Ensembl, EMBL - EBI, Cambridge, UK

15:00 - 15:40 **Viewing the Data: the UCSC browser and its possibilities**

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

15:40 - 16:20 **Coffee Break & Poster Session**

16:20 - 17:30 **PLENARY SESSION 4**

Room: Tretyakov+Morozov

16:20- 17:40 **HGVS Nomenclature: describing variants - WORKSHOP**

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

17:40 - 17:50 **Refresh Break**

17:50 - 19:15 **PLENARY SESSION 5**

Room: Tretyakov+Morozov

17:50 - 19:15 **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**

Thursday 30th May

- 8:30 - 10:30** **PLENARY SESSION 6**
Room: Tretyakov+Morozov
- 8:30 - 9:10** **Potential Consequences on the RNA Level and using prediction tools**
Andreas Laner
MGZ - Medical Genetics Centre, Munich, Germany
- 9:10 - 9:40** **Potential Consequences on Protein Level and using prediction tools**
TBA
- 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: Tretyakov+Morozov
- 11:00 - 11:30** **Copy Number Variations from Whole Exome Sequencing**
Anna Benet Pages
MGZ - Medical Genetics Centre, Munich, Germany
- 11:30 - 13:00** **HGVS, HPO and submitting Data - WORKSHOP**
Johan den Dunnen

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

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| 14:00 - 15:30 | WORKSHOP
STREAM A
Room:

ACMG classification

Andreas Laner & Anna Benet Pàges | WORKSHOP
STREAM B
Room:

Ensembl Genome Browser

Benjamin Moore |
|----------------------|--|---|

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

16:00 - 17:30

**WORKSHOP
STREAM A
Room:**

Ensembl Genome Browser
Benjamin Moore

**WORKSHOP
STREAM B
Room:**

**WES analysis using
LOVD+**
Ivo Fokkema

17:30 - 17:45

Refresh Break

17:45 - 19:15

**WORKSHOP
STREAM A
Room:**

**WES analysis using
LOVD+**
Ivo Fokkema

**WORKSHOP
STREAM B
Room:**

ACMG classification
Andreas Laner & Anna Benet Påges

19:15

END OF DAY

Friday 31 May

8:30 - 10:30 **PLENARY SESSION 8**
Room: Tretyakov+Morozov

8:30 - 9:00 **Functional Analysis of Splice Affecting Variants**

Mikhail Skoblov
Russian Center of Medical Genetics, Russian Academy of Medical Sciences, Moscow, Russia

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 **Options for evaluating variants in non-coding sequences**

Oleg Gusev
Kazan Federal University, Russia (KFU) -RIKEN "Translational genomics" Unit, RIKEN, Yokohama, Japan

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

11:00 - 12:30 **WORKSHOP
STREAM A**
Room:

UCSC Genome Browser

Robert Kuhn

**WORKSHOP
STREAM B**
Room:

Sponsored Workshop

SOFIA GENETICS

(topic TBA)

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

13:30 - 15:00 **WORKSHOP
STREAM A**
Room:

Sponsored Workshop

SOFIA GENETICS

(topic TBA)

**WORKSHOP
STREAM B**
Room:

VEP

Benjamin Moore

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

15:30 - 17:00	WORKSHOP STREAM A Room: VEP Benjamin Moore	WORKSHOP STREAM B Room: UCSC Genome Browser Robert Kuhn
17:00 - 17:10 Refresh Break		
17:10 - 18:10	PLENARY SESSION 9 Room: Tretyakov+Morozov	
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	

TO BE ALLOCATED A SESSION

NGS in practice: local Moscow lab - TBA

***WORKSHOPS** - if more workshops are arranged then the workshops on Friday 31st will become an optional choice from 3 concurrent workshops and not in a stream.