



Variant Effect Prediction Training Course

29 - 31 May 2019
Moscow, Russian Fed.

PROGRAM (29 Apr. 2019)

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

Wednesday 29th May

- 7:30 - 8:30** **REGISTRATION**
- 8:30 - 10:30** **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**
- Inna Povolotskaya
Veltischev Research and Clinical Institute of Paediatrics, Russian National Research Medical University, Russia
- 9:20 - 10:00** **NGS: what method to apply (gene panel, WES or WGS) and where technology fails**
- Ales Maver
Ljubljana University Medical Centre, UMC, Institute of Medical Genetics, Ljubljana, Slovenia
- 10:00 - 10:30** **Viewing the Data: the UCSC browser and its possibilities**
- Robert Kuhn
UCSC Genome Browser, UC Santa Cruz Genomics Institute, Santa Cruz, CA, USA

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

11:00 - 12:50 **PLENARY SESSION 2**
Room: Tretyakov+Morozov

11:00 - 11:40 **Gene Variant databases**

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

11:40 - 12:20 **Variant Classification: ACMG recommendations**

Andreas Laner
MGZ - Medical Genetics Centre, Munich, Germany

12:20 - 12:50 **Viewing the Data: the Ensembl browser and its possibilities**

Benjamin Moore
Ensembl, EMBL - EBI, Cambridge, UK

12:50 - 13:50 **Lunch**

13:50 - 16:00 **PLENARY SESSION 3**
Room: Tretyakov+Morozov

13:50 - 14:20 **Variant Annotation Integrator (VAI)**

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

14:20 - 14:50 **Variant Annotation: VEP**

Benjamin Moore
Ensembl, EMBL - EBI, Cambridge, UK

14:50 - 15:20 **Copy Number Variations from Whole Exome Sequencing**

Anna Benet Pages
MGZ - Medical Genetics Centre, Munich, Germany

15:20 - 16:00 **Potential Consequences on the RNA Level and using prediction tools**

Andreas Laner
MGZ - Medical Genetics Centre, Munich, Germany

16:00 - 16:10 **Assemble for Group Photograph**

16:10 - 16:40 **Coffee Break & Poster Session A**

16:40 - 18:10 **WORKSHOP**
STREAM A
Room: Tretyakov

Ensembl Genome Browser

Benjamin Moore

WORKSHOP
STREAM B
Room: Morozov

UCSC Genome Browser

Robert Kuhn

18:10 - 18:20 Refresh Break

18:20 - 19:20

**WORKSHOP
STREAM A
Room: Tretyakov**

**WES analysis using
LOVD+**

Ivo Fokkema

**WORKSHOP
STREAM B
Room: Morozov**

HGVS Nomenclature: describing variants

Johan den Dunnen

19.20 - 21.20 WELCOME RECEPTION

Thursday 30th May

8:30 - 9:30

**WORKSHOP
STREAM A
Room: Tretyakov**

**HGVS Nomenclature: describing
variants**

Johan den Dunnen

**WORKSHOP
STREAM B
Room: Morozov**

**WES analysis using
LOVD+**

Ivo Fokkema

9:30 - 11:00

**WORKSHOP
STREAM A
Room: Tretyakov**

UCSC Genome Browser

Robert Kuhn

**WORKSHOP
STREAM B
Room: Morozov**

Ensembl Genome Browser

Benjamin Moore

11:00 - 11.30 Coffee Break

11:30 - 13:00	<p>WORKSHOP STREAM A Room: Tretyakov</p> <p>Sponsored Workshop</p> <p>Clinical Genomics - Interpretation and Reporting with QIAGEN Bioinformatics</p> <p>Ruth Burton PhD. Clinical Applications Specialist at QIAGEN</p>	<p>WORKSHOP STREAM B Room: Morozov</p> <p>Sponsored Workshop</p> <p>SOPHiA's solutions for hereditary disorders: overcoming challenges, increasing diagnostic yield</p> <p>Boris Klyuch Business Development Manager at SOPHiA GENETICS, Nicole Grieder Subject Matter Expert at SOPHiA GENETICS, Georgios Stamoulis Clinical Application Manager at SOPHiA GENETICS & Maria Nazarenko Institute of Medical Genetics, Tomsk, Russia</p>
13:00 - 14:00 Lunch Break		
14:00 - 15:30	<p>WORKSHOP STREAM A Room: Tretyakov</p> <p>What to do with novel NGS findings (HPO, sharing, matchmaking)</p> <p>Ales Maver</p>	<p>WORKSHOP STREAM B Room: Morozov</p> <p>ACMG classification</p> <p>Andreas Laner & Anna Benet Påges</p>
15:30 - 16:00 Coffee Break & Poster Session B		
16:00 - 17:30	<p>WORKSHOP STREAM A Room: Tretyakov</p> <p>ACMG classification</p> <p>Andreas Laner & Anna Benet Påges</p>	<p>WORKSHOP STREAM B Room: Morozov</p> <p>What to do with novel NGS findings (HPO, sharing, matchmaking)</p> <p>Ales Maver</p>
17:30 - 17:45 Refresh Break		

17:45 - 19:15	WORKSHOP STREAM A Room: Tretyakov Variant Effect Predictor (VEP) Benjamin Moore	WORKSHOP STREAM B Room: Morozov Variant Annotation Integrator (VAI) Robert Kuhn
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19:15 **END OF DAY 2**
Evening at leisure

Friday 31 May

8:30 - 10:00	WORKSHOP STREAM A Room: Tretyakov Variant Annotation Integrator (VAI) Robert Kuhn	WORKSHOP STREAM B Room: Morozov Variant Effect Predictor (VEP) Benjamin Moore
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10:00 - 10:30 Coffee Break

10:30 - 12:00	WORKSHOP STREAM A Room: Tretyakov Sponsored Workshop SOPHiA's solutions for hereditary disorders: overcoming challenges, increasing diagnostic yield Boris Klyuch Business Development Manager at SOPHiA GENETICS, Nicole Grieder Subject Matter Expert at SOPHiA GENETICS, Georgios Stamoulis Clinical Application Manager at SOPHiA GENETICS & Maria Nazarenko Institute of Medical Genetics, Tomsk, Russia	WORKSHOP STREAM B Room: Morozov Sponsored Workshop Clinical Genomics - Interpretation and Reporting with QIAGEN Bioinformatics Ruth Burton PhD. Clinical Applications Specialist at QIAGEN
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12:00 - 13:00 Lunch Break

13:00 - 14:30 PLENARY SESSION 4
Room: Tretyakov+Morozov

13:00 - 14:30 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

14:30 - 15:00 Coffee Break

15:00 - 18:00 PLENARY SESSION 5
Room: Tretyakov+Morozov

15:00 - 15:30 Functional Analysis of Splice Affecting Variants

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

15:30 - 16:15 Options for evaluating variants in non-coding sequences

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

16:15 - 17:00 NGS in practice

Ekaterina Pomerantseva
"Genetico", Moscow, Russian Federation

17:00 - 17:30 Future Developments

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

17:30 - 18:00 Meeting Evaluation and Close

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

18:00 COURSE END