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, Ljublijana, 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**

16:40 - 18:10	**WORKSHOP** **STREAM A** **Room: Tretyakov**	**WORKSHOP** **STREAM B** **Room: Morozov**
	Ensembl Genome Browser Benjamin Moore	**UCSC Genome Browser** Robert Kuhn

18:10 - 18:20 **Refresh Break**

18:20 - 19:20	**WORKSHOP** **STREAM A** **Room: Tretyakov**	**WORKSHOP** **STREAM B** **Room: Morozov**
	WES analysis using LOVD+ Ivo Fokkema	**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	**WORKSHOP** **STREAM A** **Room: Tretyakov** **Sponsored Workshop** **Clinical Genomics - Interpretation and Reporting with QIAGEN Bioinformatics** Ruth Burton PhD. Clinical Applications Specialist at QIAGEN	**WORKSHOP** **STREAM B** **Room: Morozov** **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
13:00 - 14:00	**Lunch Break**	
14:00 - 15:30	**WORKSHOP** **STREAM A** **Room: Tretyakov** **What to do with novel NGS findings (HPO, sharing, matchmaking)** Ales Maver	**WORKSHOP** **STREAM B** **Room: Morozov** **ACMG classification** Andreas Laner & Anna Benet Påges

15:30 - 16:00	**Coffee Break & Poster Session**	
16:00 - 17:30	**WORKSHOP** **STREAM A** **Room: Tretyakov** **ACMG classification** Andreas Laner & Anna Benet Påges	**WORKSHOP** **STREAM B** **Room: Morozov** **What to do with novel NGS findings (HPO, sharing, matchmaking)** Ales Maver
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
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
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
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 Pâges 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**

Poster Presentations

W = Wednesday 29th May; 16:10 - 16:40
T = Thursday 30th May; 15:30 - 16:00

POS01W	Identification Of Mutations Associated With Focal Cortical Dysplasia Using Next Generation Sequencing	Vanessa Simão de Almeida
POS02T	Implementation of algorithm for reselecting reference objects for recognition methods to medical diagnosis	Nikolay Bondarenko
POS03W	Profiling the mutational spectrum of hereditary breast cancers in Turkey using multi-gene panel testing	Levent Doganay
POS04T	Novel protein-truncating variants detected in a cohort of 1,100 individuals with wide phenotypic spectrum	Aykaz Eremyan
POS05W	De novo whole exome variants in group of patients with Autism Spectrum Disorder	Yulia Grigorieva
POS06T	Disease causing variants hidden as sequencing artifacts	Hilde Tveitan Hilmarsen
POS07W	FOXG1 frameshift variant as an incidental finding: a case report.	Alina Korbut
POS08T	Prenatal diagnosis for pregnancies with abnormal ultrasound findings through Next Generation Sequencing (NGS)	Anastasia Korovko
POS09W	Mexican Node of Human Variome project: Challenges towards translational genomics	Luz Berenice López Hernández
POS10T	Mutation screening of the 81 genes by next generation sequencing (NGS) in Russian dilated cardiomyopathy patients	Vadim Mikhailov
POS11W	Preimplantation genetic testing by NGS-analysis for detection of unbalanced translocation	Elizaveta Musatova
POS12T	In-silico QA of Target Sequencing Panels	Tamara Simakova
POS13W	Genetic and phenotypic characterizations of a cohort of patients with developmental epileptic encephalopathies	Helena Tadiello de Moraes
POS14T	Rare LRRK2 gene haplotype as a potential risk factor for endemic parkinsonism	Radek Vodicka
POS15W	Efficacy of variant prediction tools on large-scale analysis using whole exome sequence in rare diseases	Kumiko Yanagi