



POCKET PROGRAM



THE
HUMAN VARIOME
PROJECT

an NGO official partner of UNESCO
sharing data · reducing disease

**31st October –
3rd November 2016**
Heraklion, Crete, Greece

**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.





Monday 31st October – The Basics

13.30 - 12.45

REGISTRATION

– Hotel Foyer

13.00 - 14.45

PLENARY SESSION 1

Room: Ariadni

Chair: Johan T. den Dunnen

13.00 - 13.15

Welcome & Introduction

Johan T. den Dunnen

Leiden Univ. Medical Center, Leiden, Netherlands

13.15 - 14.00

Variants in the genome, position and possible consequences

Jan Traeger-Synodinos

Dept. of Medical Genetics, National and Kapodistrian

University of Athens, Greece

14.00 - 14.45

Sequencing technology: Sanger, NGS and single molecule

Henk Buermans

Dept. of Human Genetics; Leiden Genome Technology Center,

Leiden, The Netherlands



14.45 - 15.15

Coffee Break

15.15 - 16.00

PLENARY SESSION 2

Room: Ariadni

Chair: Robert Kuhn

15.15 - 16.00

Calling DNA variants

Steven Laurie

CNAG (RD Connect), Barcelona, Spain

16.00 - 17.30

CONCURRENT PRACTICALS 1

Alamut (Interactive Biosoftware)

Andre Blavier & Alexandre Hatzoglou

Room: Ariadni

CONCURRENT PRACTICALS 2

VarAFT & UMD Predictor

Jean-Pierre Desvignes & Christophe Bérout

Room: Cellar

18.00 - 19.30

WELCOME RECEPTION – Astoria Capsis Hotel



Tuesday 1st November – Gathering Information

8.30 – 10.30

PLENARY SESSION 3

Room: Ariadni

Chair: Jan Traeger-Synodinos

8.30 – 9.15

DNA variants – the big databases

Robert Kuhn

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

9.15 – 9.45

Gene variant databases & sharing information

Johan den Dunnen

Leiden Univ. Medical Center, Leiden, The Netherlands

9.45 – 10.30

Human Phenotype Ontology (HPO)

Sebastian Köhler

Institute of Medical Genetics, Charité Universitätsklinikum
Berlin, Germany



10.30 – 11.00

Coffee Break

11.00 – 12.30

PLENARY SESSION 4

Room: Ariadni

Chair: Sebastian Köhler

11.00– 11.45

The Ensembl genome browser and its possibilities

Helen Sparrow

Ensembl, EMBL – EBI, Cambridge, UK

11.45 – 12.30

Prioritize: annotate and filter variants

Christophe Bérout

INSERM UMR_S910, Aix Marseille University, France

Tuesday 1st November - Gathering Information

12.30 - 14.00 Lunch Break

16.00 - 17.30

**CONCURRENT
PRACTICALS 3**

**Ensembl genome
browser**

Helen Sparrow

Room: Ariadni

**CONCURRENT
PRACTICALS 4**

**The RD-Connect
platform**

Steve Laurie

Room: Cellar

**CONCURRENT
PRACTICALS 5**

**Alamut
(Interactive
Biosoftware)**

Andre Blavier
& Alexandre
Hatzoglou

Room: Mouses



15.30 - 16.00 Coffee Break

16.00 - 17.30

**CONCURRENT
PRACTICALS 6**

**Ensembl genome
browser
(repeated)**

Helen Sparrow

Room: Ariadni

**CONCURRENT
PRACTICALS 7**

**The RD-Connect
platform
(repeated)**

Steve Laurie

Room: Cellar

**CONCURRENT
PRACTICALS 8**

**Sophia
Genetics**

Gaetano Bonifacio &
Nicole Grieder

Room: Mouses

17.30

DAY END - Evening at leisure

Wednesday 2nd November – Predicting Consequences & Variant Classification

8.30 - 10.30

PLENARY SESSION 5

Room: Ariadni

Chair: Helen Sparrow

8.30 - 9.00

Summary Session

Andreas Laner

MGZ - Medical Genetics Centre, Munich, Germany

9.00 - 9.45

Potential consequences on RNA level

Andreas Laner

MGZ - Medical Genetics Centre, Munich, Germany



9.45 - 10.30

HGVS nomenclature – describing variants

Johan den Dunnen

Leiden Univ. Medical Center, Leiden, The Netherlands

10.30 - 11.00

Coffee Break

11.00 - 12.30

PLENARY SESSION 6

Room: Ariadni

Chair: Andreas Laner

11.00 - 11.45

Variant Annotation

Helen Sparrow (EBI)

Ensembl, EMBL - EBI, Cambridge, UK

11.45 - 12.30

The UCSC genome browser and its possibilities

Robert Kuhn

UC Santa Cruz Genomics Institute

12.30 - 14.00

Lunch Break

Wednesday 2nd November – Predicting Consequences & Variant Classification

14.00 - 15.30

**CONCURRENT
PRACTICALS 9**

**UCSC genome
browser**

Robert Kuhn

Room: Ariadni

**CONCURRENT
PRACTICALS 10**

**VarAFT & UMD
Predictor
(repeated)**

Jean-Pierre
Desvignes &
Christophe Bérout

Room: Cellar

**CONCURRENT
PRACTICALS 11**

**HPO (Phenomizer)
and WES/WGS
analysis using
Exomiser**

Sebastian Köhler

Room: Mouses

15.30 - 16.00

Coffee Break

16.00 - 17.30

**CONCURRENT
PRACTICALS 12**

**UCSC genome
browser**

Robert Kuhn

Room: Ariadni

**CONCURRENT
PRACTICALS 13**

**Sophia Genetics
(repeated)**

Gaetano Bonifacio &
Nicole Grieder

Room: Cellar

**CONCURRENT
PRACTICALS 14**

**HPO (Phenomizer)
and WES/WGS
analysis using
Exomiser
(repeated)**

Sebastian Köhler

Room: Mouses

17.30 - 18.45

Free time

18.45 for 19.00

CONFERENCE DINNER @ 'Peskesi'



Thursday 3rd November – Functional Testing & Reporting

8.30 - 10.20

PLENARY SESSION 7

Room: Ariadni

Chair: Steve Laurie

8.30 – 9.15

Variant classification

Andreas Laner

MGZ - Medical Genetics Centre, Munich, Germany

9.15 - 10.20

PRESENTATIONS FROM ABSTRACTS



9.15 - 9.35

Next generation sequencing in the diagnosis of inherited cardiac disorders

Sara Benedetti

Clinical Molecular Biology Laboratory, IRCCS San Raffaele Scientific Institute, via Olgettina 60, 20132 Milano, Italy

9.35 - 9.55

Full-length CYP2D6 genotyping using PacBio RSII long reads for better drug dosage and response management

Henk P.J. Buermans

Dept. of Human Genetics; Leiden Genome Technology Center, Leiden, The Netherlands

9.55 - 10.15

Targeted RNASeq helps to improve prediction of the effect of identified variants/mutations

Bernd Dworniczak

Department of Human Genetics, University Hospital Muenster, Vesaliusweg 12, Muenster, Germany

Thursday 3rd November – Functional Testing & Reporting

10.15 - 10.20 Discussion

10.20 - 10.50 Coffee Break

PLENARY SESSION 8

Room: Ariadni

Chair: Henk Buermans

10.50 - 11.35

NGS in diagnostics

Anna Benet

MGZ - Medical Genetics Centre, Munich, Germany

11.35- 12.30

Future developments & meeting evaluation

Johan den Dunnen

Leiden Univ. Medical Center, Leiden, The Netherlands

12.30

COURSE END



WELCOME RECEPTION

Monday 31st October

6.00 pm

Welcome Reception

Astoria Capsis Hotel



CONFERENCE DINNER

Wednesday 2nd November

7.00 pm

Peskesi Restaurant

Address: Kidonias 11

behind Theotokopoulos park (El Greco park)

Short walk from the hotel, meet directly there or depart as a group

at 18.50 from the hotel lobby

The map shows the area around the Municipality of Irakleio, including the Irakleio Train Station and the Irakleio City Center. The route from the train station to the city center is marked with a blue line and a distance of 7 min 500 m. The map also shows the route from the train station to the city center, with a distance of 7 min 500 m. The map is oriented with North at the top.



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