



## *Genetic, environment and ethnicity risk factors associated with cancer in Romania*

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# SYMPOSIUM PROGRAMME

Crowne Plaza Hotel, Bucharest

Orhideea II Hall

## Monday, 24<sup>th</sup> of April, 2017

13<sup>30</sup> - 14<sup>00</sup> Registration

14<sup>00</sup> - 15<sup>00</sup> Lecture – Introduction

14:00 - 14:30 - Thorunn Rafnar - Genetic epidemiology of cancer: can it be used to reduce the burden of the disease?

14:30 - 15:00 - Paul Iordache - The impact of the ROMCAN project on Romanian public health

15<sup>00</sup> - 15<sup>20</sup> Coffee break

15<sup>20</sup> - 17<sup>00</sup> Workshop

Group work - General presentation of the workshop

## Tuesday, 25<sup>th</sup> of April, 2017

9<sup>00</sup> - 11<sup>00</sup> Lecture – Analyzing genetic data

9:00 - 9:20 - Bjarni V Halldórsson - Association study

9:20 - 9:40 - Bjarni V Halldórsson - Multiple testing

9:40 - 10:00 - Bjarni V Halldórsson - Population structure

10:00 - 10:20 - Paul Iordache - Manhattan plot and Q-Q plot

10:20 - 10:40 - Paul Iordache - Genetic loci and genes

10:40 - 11:10 - Paul Iordache - Functional classification of genetic variants

11<sup>10</sup> - 11<sup>30</sup> – Coffee break

11<sup>30</sup>-13<sup>00</sup> Workshop

11:30 - 12:00 - Group work - Genotypic data analysis

12:00 - 13:00 - Group work - Manhattan plot and Q-Q plot for expected p-values

13<sup>00</sup> - 14<sup>00</sup> Lunch

14:00 - 14:45 - Bjarni V Halldórsson, Thorunn Rafnar, Guðný A. Árnadóttir, Brynjar Ö. Jansson and Paul Iordache - Discussions regarding association studies, genetic loci and genes, multiple testing and Manhattan plot and Q-Q plot.

14:45 - 15:40 - Bjarni V Halldórsson, Thorunn Rafnar, Guðný A. Árnadóttir, Brynjar Ö. Jansson and Paul Iordache - Q&A Session

15<sup>40</sup> - 16<sup>00</sup> Coffee break

16<sup>00</sup> - 17<sup>00</sup> Discussion

### **Wednesday, 25<sup>th</sup> of April, 2017**

9<sup>00</sup> - 11<sup>00</sup> Lecture - Replication study

9:00 - 9:50 - Bjarni V Halldórsson - Genomic control

9:50 - 10:20 - Paul Iordache - Replication study

10:20 - 11:10 - Bjarni V Halldórsson - Evaluating genetic risk

11<sup>10</sup> - 11<sup>30</sup> Coffee break

11<sup>30</sup> - 13<sup>00</sup> Workshop

11:30 - 12:00 - Group work- Generating the replication data-set

12:00 - 13:00 - Group work- Manhattan plot and Q-Q plot for observed and replicated OR

13<sup>00</sup> - 14<sup>00</sup> Lunch

14:00 - 14:45 - Bjarni V Halldórsson, Thorunn Rafnar, Guðný A. Árnadóttir, Brynjar Ö. Jansson and Paul Iordache - Discussions regarding genomic control, replication studies and genetic risk

14:45 - 15:40 - Bjarni V Halldórsson, Thorunn Rafnar, Guðný A. Árnadóttir, Brynjar Ö. Jansson and Paul Iordache - Q&A Session

15<sup>40</sup> - 16<sup>00</sup> Coffee break

16<sup>00</sup> - 17<sup>00</sup> Discussion

### **Thursday, 27<sup>th</sup> of April, 2017**

9<sup>00</sup> - 11<sup>00</sup> Lecture

9:00 - 9:20 - Guðný A. Árnadóttir – Basics of whole-genome sequencing

9:20 - 9:40 - Guðný A. Árnadóttir – Basics of sequence variants and Mutations

9:40 - 10:00 - Guðný A. Árnadóttir – Frequency of sequenced variants

10:00 - 10:20 - Brynjar Ö. Jansson – Basic description of Exome Aggregation Consortium (ExAC)

10:20 - 10:40 - Brynjar Ö. Jensson – Basic description of Genome Aggregation Database (gnomAD)

10:40 - 11:10 - Brynjar Ö. Jensson – Variant - Basic description of Effect Predictor (VEP)

11<sup>10</sup> - 11<sup>30</sup> Coffee break

11<sup>30</sup> - 13<sup>00</sup> Workshop

11:30 - 11:45 - Group work - Evaluation of variants – selected examples

11:45 - 12:00 - Group work - Using online databases/tools

12:00 - 12:15 - Group work - Exemples using ExAC

12:15 - 12:30 - Group work - Exemples using gnomAD

12:30 - 12:45 - Group work - Exemples using VEP

12:45 - 13:00 - Group work- Exemples using ClinVar

13<sup>00</sup> - 14<sup>00</sup> Lunch

14:00 - 14:45 - Bjarni V Halldórsson, Thorunn Rafnar, Guðný A. Árnadóttir, Brynjar Ö. Jensson and Paul Iordache - Discussions regarding evaluation of sequenced variants.

14:45 - 15:40 - Bjarni V Halldórsson, Thorunn Rafnar, Guðný A. Árnadóttir, Brynjar Ö. Jensson and Paul Iordache - Q&A Session

15<sup>40</sup> - 16<sup>00</sup> Coffee break

16<sup>00</sup> - 17<sup>00</sup> Discussion

### **Friday, 28<sup>th</sup> of April, 2017**

9<sup>00</sup> - 11<sup>00</sup> Lecture

9:00 - 9:15 - Guðný A. Árnadóttir – Whole-genome sequencing in a clinical setting

9:15 - 9:30 - Guðný A. Árnadóttir – Mode of inheritance

9:30 - 9:45 - Guðný A. Árnadóttir – BRCA genes

9:45 - 10:00 - Guðný A. Árnadóttir – Examples of BRCA1/BRCA2 mutations in Romania

10:00 - 10:15 - Brynjar Ö. Jensson - Genotype/phenotype correlation – sources of information

10:15 - 10:30 - Brynjar Ö. Jensson - American College of Medical Genetics (ACMG) guidelines

10:30 - 10:45 - Brynjar Ö. Jensson - Online Mendelian Inheritance in Man (OMIM)

10:45 - 11:10 - Brynjar Ö. Jensson - Clinical Genomics Database (CGD)

11<sup>10</sup> - 11<sup>30</sup> Coffee break

11<sup>30</sup> - 13<sup>00</sup> Workshop

11:30 - 11:45 - Group work - Mutations in Romanian WGS data

11:45 - 12:00 - Group work - Using the tools available

12:00 - 12:15 - Group work – Frequency – Using ExAC / gnomAD

12:15 - 12:30 - Group work - Variant type and effect – Using VEP / ClinVar

12:30 - 13:00 - Group work - Genotype to phenotype – Using OMIM / CGD

13<sup>00</sup> - 14<sup>00</sup> Lunch

14:00 - 14:45 - Bjarni V Halldórsson, Thorunn Rafnar, Guðný A. Árnadóttir, Brynjar Ö. Jensson and Paul lordache - Discussions regarding Whole-genome sequencing in a clinical setting

14:45 - 15:40 - Bjarni V Halldórsson, Thorunn Rafnar, Guðný A. Árnadóttir, Brynjar Ö. Jensson and Paul lordache - Q&A Session

15<sup>40</sup> - 16<sup>00</sup> Coffee break

16<sup>00</sup> - 17<sup>00</sup> Discussion

17<sup>00</sup> – 17<sup>15</sup> End of the Symposium. Closing remarks