

Organizing committee

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Evgenia Kartsaki, Heraklion, GR
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Sotiria Kechagia, Geneva, CH
Judith Kumuthini, Cape Town, ZA
Kleanthi Lakiotaki, Heraklion, GR
Ming Ta Michael Lee, Yokohama, JP
Milan Macek Jr, Prague, CZ
Max van Min, Utrecht, NL
Clint Mizzi, Msida, MT
George P. Patrinos, Patras, GR
George Potamias, Heraklion, GR
Olaf Riess, Tübingen, DE
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Alessio Squassina, Cagliari, IT
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Pharmacogenomics
and Genomic Medicine:
Bridging Research
and the Clinic



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Pharmacogenomics
and Genomic Medicine:
Bridging Research and the Clinic

September
11 - 15
2014

Apollo Resort hotel
Aegina island,
GREECE

Scientific Program

SCIENTIFIC PROGRAM

Day 1 - Thursday, 11 September 2014

08:00 – 14:30 Arrivals AND Delegates registration

SESSION 1 – Chairperson: G. Patrinos

14:30 – 14:45 2014 Golden Helix Summer School coordinators
Introduction

14:45 – 15:15 George P. Patrinos (Patras, Greece)
Genomic Medicine in the global village

15:15 – 15:45 Emmanouil Dermitzakis (Geneva, Switzerland)
From population and personalized genomics to personalized medicine

15:45 – 16:15 Coffee break

16:15 – 17:30 WORKSHOP 1 – RD-CONNECT
A. Milan Macek (Prague, Czech Republic)
European genetics and rare disease initiatives
B. Ivo Gut (Barcelona, Spain)
RD-Connect: A database for rare disorders research
C. Olaf Riess (Tubingen, Germany)
RD-Neuomics: -omics research for diagnosis and therapy in rare neuromuscular and neurodegenerative diseases: An EU-funded FP7 project

17:30 – 18:15 Poster Presentations 1 (PO.01-PO.07)

18:15 – 19:30 DAY ENDS - FREE TIME

19:30 – 22:30 Welcome reception

Day 2 – Friday, 12 September 2014

08:00 – 09:30 Breakfast

09:30 – 11:00 Flash Talks 1 – Chairperson: S. Pavlovic

FT.01. Vesna Spasovski (Belgrade, Serbia)
The first extracellular loop of the PTCH1 protein: a true “hot spot” for PTCH1 gene?
FT.02. Irena Glumac (Belgrade, Serbia)
High WT1 expression as a new molecular marker of poor prognosis in Serbian Acute Myeloid Leukemia patients with normal karyotype (AML-NK)
FT.03. Danijela Drakulic (Belgrade, Serbia)
Characterization of the 22q11.2 region in patients with clinical features of 22q11.2 deletion syndrome
FT.04. Nataša Kovačević-Grujičić (Belgrade, Serbia)
Antitumor effect of Meripilus giganteus Karst. methanolic extract via upregulation of p53, BAX and SOX1 expression in HeLa cells
FT.05. Danijela Stanisavljevic (Belgrade, Serbia)
SOX14 expression during retinoic acid induced neural differentiation of pluripotent embryonal carcinoma stem cells
FT.06. Adrijana Klajn (Belgrade, Serbia)
SOX2 overexpression affects neural differentiation of human pluripotent NT2/D1 cells
FT.07. Milena Milivojevic (Belgrade, Serbia)
Functional characterisation of novel dominant-negative mutant of the human SOX18 protein
FT.08. Biljana Stankovic (Belgrade, Serbia)
Inflammatory bowel disease in Serbian population: association of proinflammatory gene variants with the disease and their combined contribution in the prediction of disease development
FT.09. Natasa Tomic (Belgrade, Serbia)
Molecular genetic markers as a base for administration of targeted therapy in acute promyocytic leukemia

SESSION 2 – Chairperson: B. Ylstra

11:00 – 11:30 Alessio Squassina (Cagliari, Italy)
Genomics and microarray technology

11:30 – 12:00 Bauke Ylstra (Amsterdam, the Netherlands)
Spatial copy number heterogeneity and relevance of chromosome 10q deletion in low-grade gliomas by whole genome sequencing

12:00 – 12:30 Nduna Dzimiri (Riyadh, Saudi Arabia)
The genetic intricacy of treating coronary artery disease

12:30 – 14:00 Lunch break

SESSION 3 – Chairperson: E. Dermitzakis

14:00 – 14:30 Ming Ta Michael Lee (Yokohama, Japan)
Pharmacogenomics and personalized medicine: From basic research to clinical application

14:30 – 15:00 Ron H. van Schaik (Rotterdam, the Netherlands)
Clinical pharmacogenomics and genomic medicine

15:00 – 15:30 Vita Dolzan (Ljubljana, Slovenia)
Clinical pharmacogenetic models for translation of pharmacogenetics to clinical practice

15:30 – 16:00 Branka Zukic (Belgrade, Serbia)
Pharmacogenomics of 6-mercaptopurin treatment in childhood acute lymphoblastic leukemia

16:00 – 16:30 Coffee break

SESSION 4 – Chairperson: E. Vayena

16:30 – 17:00 Effy Vayena (Zurich, Switzerland)
Personal Genomics: Clinical and personal utility at an ethical crossroad

17:00 – 17:30 Sotiria Kechagia (Geneva, Switzerland)
Direct-to-Consumer genetic and pharmacogenomic testing: A legal perspective

17:30 – 18:15 Poster Presentations 2 (PO.08-PO.14)

18:15 – 19:30 DAY ENDS - FREE TIME

19:30 – 21:00 Dinner

Day 3 - Saturday, 13 September 2014

08:00 – 09:30 Breakfast

09:30 – 11:00 Flash Talks 2 – Chairperson: P. Danielson

FT.10. Philip Danielson (Denver, CO, USA)
Application of Mass Spectrometry in Forensic Serology
FT.11. Magdalena Stepien (Zurich, Switzerland)
A physician’s perspective on direct-to-consumer genetic testing
FT.12. Nantia Flora (Patras, Greece)
Genetic variation as a predictor of smoking behavior
FT.13. Aldesia Provenzano (Florence, Italy)
Heterogeneous genetic alterations in sporadic nephrotic syndrome associate with resistance to immunosuppression.
FT.14. Sabrina Giglio (Florence, Italy)
Therapeutic implications of novel mutations of RFX6 gene associated with early-onset diabetes
FT.15. Jihène Bouassida (Tunis, Tunisia)
Screening of CYP1B1 gene in Tunisian patients with Primary Congenital Glaucoma
FT.16. Konstantina Chalikiopoulou (Patras, Greece)
Pharmaco-epigenomic analysis of MAP3K5 gene promoter in β -type hemoglobinopathies patients under hydroxyurea treatment
FT.17. Vasiliki Chondrou (Patras, Greece)
Differential molecular profiling of human hematopoietic tissues during human ontogenesis - A hemoglobinopathies therapeutics approach
FT.18. Eleni Dalabira (Patras, Greece)
Pharmacogenomics in Europe: Applications in public health

SESSION 5 – Chairperson: A. Squassina

11:00 – 11:30 Clint Mizzi (Msida, Malta)
Whole genome sequencing and data interpretation in genomic medicine

11:30 – 12:00 George Potamias (Heraklion, Greece)
Boosting pharmacogenomics: A translational bioinformatics perspective

12:00 – 12:30 George P. Patrinos (Patras, Greece)
Databases in genomics

12:30 – 14:00 Lunch break

SESSION 6 – Chairperson: G. Potamias

14:00 – 14:45 CORPORATE LECTURE – CERGENTIS
Max van Min (Utrecht, the Netherlands)
Targeted locus amplification

14:45 – 15:15 Judit Kumuthini (Cape Town, ZA)
Sustainable capacity development in personalized medicine

15:15 – 15:45 Athanassios Vozikis (Piraeus, Greece)
The intersection of genomics and health economics

15:45 – 16:15 Coffee break (sponsored by eMoDiA)

16:15 – 17:30 WORKSHOP 2 – eMoDiA
A. Theodora Katsila (Patras, Greece)
Pharmacogenomics Initiatives : eMoDiA
B. George Potamias (Heraklion, Greece)
Towards an electronic PharmacoGenomics Assistant (ePGA)
C. Evgenia Kartsaki (Heraklion, Greece)
Pharmacogenomic Translation Services
D. Kleanthi Lakiotaki (Heraklion, Greece)
A Pharmacogenomics Dashboard of 1000 Genomes Project

17:30 – 19:30 DAY ENDS – FREE TIME

19:30 – 21:00 Dinner (sponsored by eMoDiA)

Day 4 – Sunday, 14 September 2014

08:00 – 09:00 Breakfast

09:00 – 12:30 Visit at the AFEA archaeological site and museum

12:30 – 14:30 Lunch break at Aghia Marina

SESSION 7 – Chairperson: M. Ferrari

14:30 – 15:00 Maurizio Ferrari (Milan, Italy)
How to improve molecular diagnostic literacy in developing countries

15:00 – 15:30 George P. Patrinos (Patras, Greece)
Public Health Genomics: From concept to reality

15:30 – 16:00 Coffee break

16:00 – 17:30 Flash Talks 3 – Chairperson: T. Katsila

FT.19. Cristiana Pavlidis (Patras, Greece)
Whole genome sequencing analysis of a Greek family affected by Adamantiades-Behcet disease
FT.20. Eleni Merkouri-Papadima (Patras, Greece)
Identification of genomic variants leading to Amyotrophic Lateral Sclerosis Causes in Greek population using a whole genome sequencing approach
FT.21. Katja Goričar (Ljubljana, Slovenia)
Clinical-pharmacogenetic model for prediction of treatment outcome in malignant mesothelioma
FT.22. Barbara Jenko (Ljubljana, Slovenia)
The pharmacogenetic model of influence of transporters polymorphisms and clinical factors on methotrexate inefficacy in rheumatoid arthritis patients
FT.23. Maria Manica-Cattani (Santa Maria, Brazil)
Guaraná and its main bioactive molecules modulates different
FT.24. Kleanthi Lakiotaki (Heraklion, Greece)
Towards a pharmacogenomics information system for personalized medicine
FT.25. Evangelia-Eirini Tsermpini (Patras, Greece)
Role of the rs6313 and rs1799978 variants as pharmacogenomic biomarkers in Greek, Italian and Croatian schizophrenia patients
FT.26. Konstantinos Siatis (Patras, Greece)
Development of a novel tumor homing compound targeting selectively glioblastoma cells
FT.27. Zoi Zagoriti (Patras, Greece)
Assessment of the STAT4 rs7574865 SNP in genetic susceptibility of early-onset Myasthenia gravis

17:30 – 18:15 Poster Presentations 3 (PO.15-PO.21)

18:15 – 19:30 DAY ENDS - FREE TIME

19:30 – 22:00 Farewell Dinner – Traditional Party

Day 5 – Monday 15 September 2014

08:00 – 09:30 Breakfast

09:30 – 10:45 Career development session
Vita Dolzan (Ljubljana, SI; Chairperson)
Sonja Pavlovic (Belgrade, RS)
Emmanouil Dermitzakis (Geneva, CH)
Judit Kumuthini (Cape Town, ZA)
A. Nazli Basak (Istanbul, TR)
Philip Danielson (Denver, CO, USA)
Nduna Dzimiri (Riyadh, SA)

THE EMBO LECTURE – Chairperson: G. Patrinos

10:50 – 11:00 Introduction
11:00 – 11:45 Douglas R. Higgs (EMBO Member, Oxford, UK)
Switching genes on and off during differentiation and development

BEST ABSTRACTS SESSION – Chairperson: G. Patrinos

11:45 – 12:00 Best Abstract 1 – M Bartsakoulia, V Boczonadi, A Gomez-Duran, P Yu W Man, P F Chinnery, R Horvath (Newcastle, UK)
Studying the effect of L-cysteine in MELAS and MERRF

12:00 – 12:15 Best Abstract 2 – S Pavlovic, B Stanic, J Kostic, N Pejanovic, B Lucic, N Tomic, I Glumac, D Janic, L Dokmanovic, S Jankovic, N Suvajdzic Vukovic, D Tomin, B Mihaljevic, M Todorovic, J Jelcic, D Krstajic, M Popovic, I Bogicevic, M Stojiljkovic (Belgrade, Serbia)
Next-generation sequencing analysis of hematological malignancies: Serbian experience

12:15 – 12:30 2014 Golden Helix Summer School coordinators
Conclusions

12:30 Delegates departure