

Invited speakers and Chairmen

Nigel Carter (Hinxton, UK)
 Jean-Jacques Cassiman (Leuven, BE)
 Gianpiero Cavalleri (Dublin, IRL)
 George Chrousos (Athens, GR)
 Nancy Cox (Chicago, IL, USA)
 Johan den Dunnen (Leiden, NL)
 Radoje Drmanac (Mountain View, CA, USA)
 Beverly Emanuel (Philadelphia, PA, USA)
 Paolo Fortina (Philadelphia, PA, USA)
 Frank Grosveld (Rotterdam, NL)
 Federico Innocenti (Chicago, IL, USA)
 Thomas Joos (Tubingen, DE)
 Manolis Kanavakis (Athens, GR)
 Sandra Kirkwood-Close (USA)
 Eric Knudsen (Philadelphia, PA, USA)
 Larry Kricka (Philadelphia, PA, USA)
 Cedric le Caignec (Nantes, FR)
 Kevin Long (Chapel Hill, NC, USA)
 Lucio Luzzatto (Toscane, IT)
 Joannis Mytilineos (Ulm, DE)
 Philippos Patsalis (Nicosia, CY)
 George P. Patrinos (Patras, GR)
 Michael B. Petersen (Athens, GR)
 Christian Stoeckert (Philadelphia, PA, USA)
 Graham Taylor (Leeds, UK)
 Jan Traeger-Synodinos (Athens, GR)
 Athanassios Tsiatis (Baltimore, MD, USA)
 Ron van Schaik (Rotterdam, NL)
 Bauke Ylstra (Amsterdam, NL)



Genetic Analysis In Translational Medicine



Genetic Analysis In Translational Medicine

Scientific Program

Founder



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COMMUNICATION



December
1st - 4th
2010

Under the auspices of
His Excellency the President of the Hellenic Republic
Dr. Karolos Papoulias

DIVANI Acropolis
Palace hotel
Athens, Greece



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We are very pleased to welcome you to the 2010 **GOLDEN HELIX Symposium®**:
"Genetic Analysis in Translational Medicine".

Recognizing the challenges that currently emerge for molecular medicine in the post-genomic era, the **GOLDEN HELIX Symposium®** on Genetic Analysis and Translational Medicine includes lectures by more than 20 internationally renowned scientists in the field.

The scientific program covers all aspects of this discipline, including Genome-wide Association Studies, Genomics of cancer and human disorders, Molecular Cytogenetics, Genomic technology, next-generation sequencing, Pharmacogenomics, Bioinformatics, Genetics and Society and Genetic analysis in clinical practice.

We are committed to setting the highest standard symposia in the field of human genomics and personalised medicine, and look forward to a very successful meeting.

With best regards,

George P. Patrinos Department of Pharmacy, University of Patras School of Health Sciences, Rion, Patras, Greece	Federico Innocenti Department of Medicine, University of Chicago Chicago, IL, United States of America	Paolo Fortina Kimmel Cancer Center Thomas Jefferson University Philadelphia, PA, United States of America	Nancy Cox Department of Medicine, University of Chicago Chicago, IL, United States of America
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SCIENTIFIC PROGRAM

Wednesday, December 1st, 2010

15:00-19:30 DELEGATE REGISTRATION
20:00 WELCOME COCKTAIL

Thursday, December 2nd, 2010

08:30-08:45 Opening addresses
George P. Patrinos, Organizing committee, University of Patras, Greece
Dimitrios Kalpaxis, Vice Rector, University of Patras, Greece
Stavros Besbeas, President, Hellenic Anticancer Society, Greece

SESSION 1 – OMICS
(Chairpersons: G. Chrousos, E. Kanavakis)

08:45-09:15 **Erik Knudsen (Philadelphia, PA, USA)**
Transcription and microRNA profiling

09:15-09:45 **Nancy Cox (Chicago, IL, USA)**
Genome-wide association studies for complex traits

09:45-10:30 **1st Keynote lecture**
Jean-Jaques Cassiman (Leuven, BE)
Policies and guidelines for genetic testing

10:30-10:45 COFFEE BREAK

10:45-11:15 **George P. Patrinos (Patras, GR)**
Unraveling the molecular mechanism of human fetal globin gene silencing

11:15-11:45 **Gianpiero Cavalleri (Dublin, IE)**
Genetic mapping in epilepsy predisposition and treatment

11:45-12:15 **Larry Kricka (Philadelphia, PA, USA)**
Consumer Genomics

12:15-12:45 **Kevin Long (Chapel Hill, NC, USA)**
Population-ized medicine: The PGENI project

12:45-14:00 LUNCH BREAK

14:00-14:30 **COMPANY LECTURES – I**
Mark Ross (Illumina)
Characterizing somatic mutations in cancer genomes using Illumina sequencing

14:30-14:50 **Marco L. Pirotta (Life Technologies)**
Advances in next-generation sequencing

14:50-16:00 COFFEE BREAK - POSTER SESSION I

SESSION 2 – Molecular Cytogenetics
(Chairpersons: B. Ylstra, N. Cox)

16:00-16:30 **Nigel Carter (Hinxton, UK)**
Molecular Cytogenetics

16:30-17:00 **Bauke Ylstra (Amsterdam, NL)**
Molecular cytogenetics in oncology: From technique to therapeutic target

17:00-17:30 **Beverly Emanuel (Philadelphia, PA, USA)**
Copy Number Variation: cause and susceptibility factor for recurrent genomic disorders

17:30-18:00 **Cedric Le Caignec (Nantes, FR)**
Application of oligoarray CGH in the diagnostic genetic laboratory

20:00 WINE TASTING EVENT (By invitation only)

Friday, December 3rd, 2010

08:30-08:45 Opening
Paolo Fortina, Organizing committee, Thomas Jefferson University, Philadelphia, PA, USA

SESSION 3 – Next generation sequencing applications
(Chairpersons: P. Fortina, G. Patrinos)

08:45-09:15 **Paolo Fortina (Philadelphia, PA, USA)**
Overview of next-generation sequencing technologies

09:15-09:45 **Frank Grosveld (Rotterdam, NL)**
Next generation sequencing application in the study of transcription

09:45-10:30 **2nd Keynote lecture**
Lucio Luzzatto (Florence, IT)
The rate of somatic mutations and the development of cancer

10:30-10:45 COFFEE BREAK

10:45-11:15 **Radoje Drmanac (Mountain View, CA, USA)**
Next-generation sequencing applications: Towards a 1000\$ genome

11:15-11:45 **Graham Taylor (Leeds, UK)**
Next-generation DNA sequencing in the clinic

11:45-12:15 **Christian Stoeckert (Philadelphia, PA, USA)**
Bioinformatics: Making sense of genomics data

12:15-12:45 **Johan den Dunnen (Leiden, NL)**
Pathogenic or not - the value of gene variant databases

12:45-14:00 LUNCH BREAK – WORKSHOP (Sponsored by Agilent Technologies)

COMPANY LECTURES – II

14:00-14:30 **Maria Tzeti (Agilent Technologies)**
Array CGH in a clinical setting

14:30-14:50 **Rick Hockett (Affymetrix)**
The DMET platform

14:50-16:00 COFFEE BREAK - POSTER SESSION II

SESSION 4 – Current genetic technology in diagnostics and therapy
(Chairpersons: J. den Dunnen, G. Taylor)

16:00-16:30 **Johan den Dunnen (Leiden, NL)**
High resolution melting curve analysis

16:30-17:00 **Athanassios Tsiatis (Baltimore, MD, USA)**
Pyrosequencing: Application to cancer diagnostics and epigenetic studies

17:00-17:30 **Thomas Joos (Tubingen, DE)**
Application of proteomics in disease diagnostics

17:30-18:00 **Aglaia Athanassiadou (Patras, GR)**
Episomal DNA as vector for gene therapy

Evening free

Saturday, December 4th, 2010

08:30-08:45 Opening
Federico Innocenti, Organizing committee, University of Chicago, Chicago, IL, USA

SESSION 5 – Translating genetic analysis into clinical practice
(Chairpersons: F. Innocenti, R. van Schaik)

08:45-09:15 **Ron van Schaik (Rotterdam, NL)**
Implementation of Pharmacogenetics in clinical diagnostics: Results from 5 year experience

09:15-09:45 **Sandra Kirkwood-Close (USA)**
Pharmacogenomics clinical trials

09:45-10:15 **Philippos Patsalis (Nicosia, CY)**
FoSTeS/MMBIR and NAHR at the human proximal Xp region and the mechanisms of human Xq isochromosome formation

10:15-10:30 COFFEE BREAK

COMPANY LECTURE – III

10:30-11:00 **Ruth Burton (Oxford Gene Technology)** The latest developments in aCGH from OGT

11:00-11:30 **Joannis Mytilineos (Ulm, DE)**
Immunogenetics

11:30-12:00 **Jan Traeger-Synodinos (Athens, GR)**
Prenatal, non-invasive and pre-implantation genetic diagnosis of inherited disorders: Lessons from hemoglobinopathies

12:00-12:30 **Michael B. Petersen (Athens GR)**
Prognostic markers for complex diseases: The paradigm for non-syndromic deafness

Best Posters Presentation

12:30-12:45 Poster I: Mike Makrigiorgos (Boston, MA, USA)

12:45-13:00 Poster II: Marianna Bei (Charlestown, MA, USA)

13:00-13:15 Closing Remarks and Future Meetings

MEETING ENDS