



# IPOKRATES

## Clinical Seminar

Foundation

## Applied Clinical Genomics

Kifisia, Athens, Greece  
April 23-25, 2018

### Objectives

IPOKRATES seminars provide high quality postgraduate education, which enable professionals to keep abreast of the most recent developments and offer participants the opportunity to discuss clinical problems or scientific issues personally with international experts. This seminar is designed to provide the researchers and clinicians to present the most recent developments, acquisitions and the broad evolution of Clinical Genetics to Clinical Genomics, therefore allowing a broad and in-depth discussion on this modern tool in clinical practice.

### Lecturers

Jill Clayton-Smith	MBBCh,MD,FRCP, Honorary Professor, Division of Evolution & Genomic Sciences, Manchester University and Manchester University Foundation Trust, England
Bartsocas, Christos	Emeritus Professor of Pediatrics, University of Athens, Consultant Athens Pediatric Center, Athens, Greece
Bruno Dallapiccola	MD, Professor, Direttore Scientifico Ospedale Pediatrico Bambino Gesù,IRCCS, Rome, Italy
Sofia Douzgou	MD,PhD,FRCP, Honorary Senior Lecturer, Division of Evolution & Genomic Sciences, Manchester University and Manchester University Foundation Trust, England
Sophia Kitsiou-Tzeli	MD,PhD, Professor of Medical Genetics, University of Athens Medical School, Athens, Greece
Peter M.Krawitz	MD,PhD, Head, Institute for Genomic Statistics & Bioinformatics University of Bonn, Germany
Periklis Makrythanasis	MD,PhD,FMH,PD, Clinical Geneticist, Biomedical Research Foundation of the Academy of Athens
Gijs Santen	MD,PhD, Clinical Geneticist, Leiden University Medical Center, Leiden, The Netherlands
Aspasia Tsezou	Professor of Medical Genetics, University of Thessaly Medical School, Larissa, Greece

Limited Number of Participants: 60

Please Note: Only registered participants, NO on site registration!

Please register via internet: <http://www.ipokrates.info>

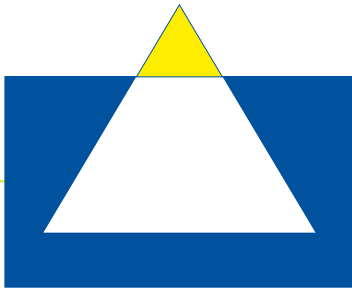
**Program Design** Christos S. Bartsocas, University of Athens, Greece  
Sofia Douzgou, Manchester University Foundation NHS Trust

**Target Groups** Physicians, Geneticists, Biologists from academic and non-university centers, hospitals and laboratories. This includes Pediatricians and Clinical Geneticists dealing with the care of patients with inherited and particularly rare diseases.

**Major Sponsors** Dräger, Elsevier, Nestlé Nutrition Institute

**Local Sponsors** Athens Medical Group, Pharmaserve-Lilly, Sanofi-Genzyme, MENARINI Pharmaceutical Company, FDNA





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## Local Host

**Christos S. Bartsocas, MD Emeritus Professor of Pediatrics**

Consultant, Athens Pediatric Center, email: [cbartsocas@nurs.uoa.gr](mailto:cbartsocas@nurs.uoa.gr)  
Sofia Douzgou, Manchester, UK, email: [sofia.douzgou@mft.nhs.uk](mailto:sofia.douzgou@mft.nhs.uk)

## Information/ Registration

**IPOKRATES Head Office c/o m:con – mannheim:congress GmbH**

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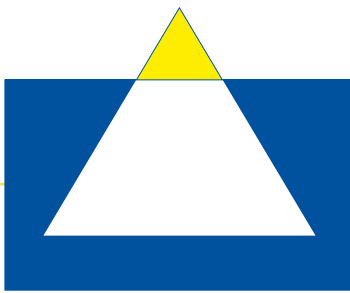
## Travel Visa

If Visa is required, each participant is responsible for his/her VISA, and IPOKRATES assumes NO liability

## Contributions to Cover Expenses

Participation Fees	Doctors of hosting country	Doctors from outside
	€ 150.-	€ 300.-

**Subject to change.**



# Applied Clinical Genomics

Kifissia, Greece  
April 23-25, 2018

## Monday, April 23, 2018

09.00-09.30 *Registration - Coffee*  
09.30-10.00 Welcome addresses

Christos S.Bartsocas, Sofia Douzgou and  
Nikos Moschos (Athens Medical Center)  
C.S.Bartsocas  
Jill Clayton- Smith

10.00-10.30 A tribute to Victor A.McKusick  
10.30-11.30 The story of the Deciphering Developmental Disorders Project  
11.30-11.45 *Coffee Break*  
11.45-12.45 Clinical Interpretation of NGS data  
12.45-13.45 Crowdsourcing for rare dysmorphic syndromes  
13.45-15.00 *Lunch break (light lunch)*  
15.00-16.00 Facial recognition technology  
16.00-17.00 Genomics of Osteoarthritis

Gijs Santen  
Sofia Douzgou

Peter Krawitz  
Aspasia Tsezou

## Tuesday, April 24, 2018

09.00-10.00 Whole Exome Sequencing outcomes in "undiagnosed" patients  
10.00-11.00 PIK3CA phenotypes  
11.00-11.15 *Coffee Break*  
11.15-12.15 Genetics of spliceosomal disorders  
12.15-13.15 Mabry syndrome  
13.15-14.30 *Lunch Break (light lunch)*  
14.30-15.30 The Bambino Gesù hospital experience on undiagnosed patients:  
clinical and NGS results  
15.30-16.30 KAT6B phenotypes  
16.30-17.30 Ethical issues in the practice of Clinical Genetics

Bruno Dallapiccola  
Jill Clayton - Smith

Sofia Douzgou  
Peter Krawitz

Bruno Dallapiccola

Jill Clayton- Smith  
Periklis Makrythanasis

## Wednesday, April 25, 2018

09.00- 10.00 MEF2C haploinsufficiency syndrome as a paradigm of Rett-like syndromes  
10.00-11.00 Prenatal and Neonatal WES  
11.00-11.15 *Coffee Break*  
11.15-12.15 Genetic and Genomic Counselling  
12.15-13.15 ARID1B experiences: phenotypes and startup of a multidisciplinary clinic  
13.15 Summing - up and end of the Seminar

Sofia Douzgou  
Gijs Santen

Sophia Kitsiou  
Gijs Santen

**Subject to change.**