



Organised by:

ΕΛΛΗΝΙΚΗ ΕΤΑΙΡΕΙΑ ΙΑΤΡΙΚΗΣ ΓΕΝΕΤΙΚΗΣ
HELLENIC SOCIETY OF MEDICAL GENETICS



National and Kapodistrian
University of Athens
UNIVERSITY SCHOOL OF MEDICINE
DEPARTMENT OF MEDICAL GENETICS

“Shaping the future
of Clinical Genetics
through
Innovative
Technologies”



SYMPOSIUM in Clinical Genetics & Genomics

with international participation

18th Educational Seminar in Genetics

1&2 June 2018
Aegli Zappiou | Athens

PRELIMINARY PROGRAM

www.genetics2018.mdcongress.gr

Symposium in Clinical Genetics and Genomics

18th Educational Seminar in Genetics

Welcome from the President

Dear colleagues and friends,

The Department of Medical Genetics of the National and Kapodistrian University of Athens, School of Medicine and the Hellenic Society of Medical Genetics would like to welcome you to the Symposium in Clinical Genetics and Genomics with International speakers from the USA, Europe and Greece. The Symposium will take place on the 1st and 2nd June 2018, in **Aegli Zappiou**, Athens (Greece) entitled: "**Shaping the Future of Clinical Genetics through Innovative Technologies**".

The aims and objectives of the ERASMUS+project's (NGENES) as well as the intellectual outputs during the Symposium are going to be presented in order to gain the maximum benefits of the project. The goal is to achieve a high rate of participation to maximize the dissemination impact and to acquire knowledge on the innovative outcomes. The exchange of knowledge and the discussion on the findings are going to be taken into account for the evaluation process.

The 2018 Symposium sessions will explore the following topics:

- Neuromuscular Genomics
- Cancer Genomics
- Genetics of Skeletal Dysplasias
- Genetics of Renal Diseases
- Neurodevelopmental Genomics
- Mitochondrial Disorders and Metabolic Disorders
- Prenatal Diagnosis

The aim and expectations of this Symposium is to bring together Medical and Clinical Scientists, Researchers, Molecular Biologists, other Health Care Professionals and Industry. Also, empowering clinicians with the information they require to intergrate genomics into medical practice. It will provide a unique opportunity for sharing scientific knowledge in the evolving field of Precision Medicine / New Clinical Genomics and facilitate networking collaborations.

My sincere thanks for your support in this very significant effort at the time of the official recognition of the speciality of Medical Genetics and Laboratory Genetics in Greece.

On behalf of the Organizing and the Scientific Committee

Kind Regards

The President of the Organizing Committee

Helena Fryssira, MD, MSc, PhD

Associate Professor in Clinical Genetics

Head of Medical Genetics Department,

National and Kapodistrian University of Athens, Greece

ORGANIZING COMMITTEE

President of the Organizing Committee

Fryssira Helena

Members

Amenta Styliani	Poulou Myrto
Kakourou Georgia	Psoni Stavroula
Kekou Kyriaki	Selenti Nikoleta
Kokkinis Charis	Sofocleous Christalena
Kolialexi Aggeliki	Tsaroucha Charoula
Kosma Konstantina	Tsoutsou Eirini
Labrakis Antonis	Vrettou Christina

INTERNATIONAL SCIENTIFIC COMMITTEE

Chiurazzi Pietro	Mercuri Eugenio
Deltas Constantinos	Mortier Geert
Gorman Grainne Siobhan	Ramos Feliciano J.
Hurst Jane	Rolfs Arndt
Katsanis Nicholas	Sangiorgi Luca
Lochmuller Hanns	

SCIENTIFIC COMMITTEE

Anastasopoulos Ioannis	Mastorakos George
Antonarakis Stylianos	Mavrou Ariadni
Antsaklis Aristides	Metaxotou Aikaterini
Athanasiadis Apostolos	Moschonas Nikolaos
Bacopoulou Flora	Moschovi Maria
Bartsokas Christos	Pampanos Andreas
Charmandari Evangelia	Pangalos Konstantinos
Chrousos George	Papadimas George
Creatsas George	Papaevangelou Vana
Deligeoroglou Efthimios	Papaggelopoulos Panos
Drogari Euridiki	Papantoniou Nikolaos
Eleftheriades Makarios	Papavasiliou Antigoni
Fidani Styliani	Pervanidou Neni
Georgiou Ioannis	Petersen Michael
Giouroukos Sotirios	Pons Roser
Gyftodimou Giolanta	Savidou Olga
Kanaka-Gantenbein Christina	Sifakis Stavros
Kanavakis Emmanuel	Stefanis Leonidas
Kattamis Antonios	Synodinos-Traeger Joanne
Kattamis Christos	Tsezou Aspasia
Kitsiou-Tzeli Sofia	Tsolia Maria
Kosmaidou-Aravidou Zoe	Tzetzis Maria
Loutradis Dimitrios	Tzoumaka-Bakoula Chrysa

FRIDAY
JUNE 1st
2018

- 09:30-19:30 **Registrations**
- 10:00-10:30 **Welcome Addresses**
Introduction
Helena Fryssira
- 10:30-11:30 **1st Section: Metabolic and Mitochondrial disorders**
Chairs: Bartsocas C., Mastroianni S.
Mitochondrial disorders
Graine Siobhan Gorman

Early diagnosis of Inborn Errors of Metabolism based on Clinical
and Biochemical Criteria
Euridiki Drogari
- 11:30-12:00 **Coffee break**
- 12:00-13:00 **Plenary lectures**
Chairs: Metaxotou A., Kanavakis E.
Evolution vs Development: Genetic Disorders of the Epigenetic Machinery
George Chrousos

Collagen IV Nephropathy as a Phenotypic Chameleon: How a
Monogenic disease behaves as a Complex disorder
Constantinos Deltas
- 13:00 -14:00 **Plenary Lectures : Neuromuscular disorders**
Chairs: Skouteli E., Papadimas G.
Therapeutic advances in DMD and SMA
Eugenio Mercuri

Inherited disorders of neuromuscular transmission – diagnosis and
personalized therapies
Hanns Lochmuller
- 14:00-15:00 **Break**

FRIDAY
JUNE 1st
2018

Scientific Program

15:00-17:00

2nd Section: Prenatal genetics

Chairs: Sifakis S., Kolialexi A., Amenta S.

Prenatal diagnosis of fetal abnormalities. Advances and limitations
Apostolos Athanasiadis

Intrauterine Fetal Surgery
Nikolaos Papantoniou

Non-invasive Prenatal Screening-NIPS
Ariadni Mavrou

Genetic Counselling
Sofia Kitsiou-Tzeli

17:00-18:30

Plenary lectures

Chairs: Moschonas N., Traeger-Synodinos J.

Rare Diseases: The use of new technologies in the diagnosis of congenital malformation syndromes and intellectual disability
Jane Hurst

Reverse Genetics – the Example of Congenital Deafness
Michael Petersen

18:30-19:30

**Opening Ceremony
Greetings**

Meropi Tzoufi

State Secretary for Education Research and Religious Affairs

19:30

WELCOME DRINK

- 09:30-17:00 **Registrations**
- 10:00:11:00 **3rd Section: Cancer Genomics**
Chairs: Charmandari E., Tzetis M.
Genomics in Pediatric Hematology-Oncology
Antonios Kattamis

Genetic abnormalities may alter the therapeutic decision
Maria Moschovi
- 11:00-12:00 **Plenary Lectures**
Chairs: Pampanos A., Sofocleous C.
X-Linked Intellectual Disability / Mental Retardation
Pietro Chiurazzi

Cohesinopathies
Feliciano Ramos
- 12:00-12:30 **Coffee break**
- 12:30-14:00 **4th Section: Neurodevelopmental disorders**
Chairs: Bacoula C., Fidani L., Tsoutsou E.
Classic congenital Adrenal Hyperplasia
Evangelia Charmandari

The state of the art investigation of short stature
Christina Kanaka-Gantenbein

Persistent Organic Pollutans (POPs) in children with
Neurodevelopmental disorders
Neni Pervanidou
- 14:00-15:30 **Break**
- 15:30-16:00 **Plenary lecture**
Chairs: Mavrou A., Kolialexi A.
Importance of proteomics in rare hereditary diseases
Arndt Rolfs
- 16:00-17:30 **Plenary lectures**
Chairs: Fryssira H., Anastasopoulos I., Savvidou O.
Genetics of Skeletal Dysplasias (ERASMUS+PROJECT-NGENES)
Geert Mortier
Luca Sangiorgi
- 17:30 **Discussion - Closing of the Symposium**

ABSTRACTS SUBMISSION GUIDELINES FOR E-POSTERS

1. Abstracts submitted via fax or via regular mail will NOT be accepted.
2. Abstracts should be submitted in English only. IMPORTANT! Any person submitting an abstract can only be the first author for two abstracts. Registration to the meeting is required for the first author of each abstract.
3. Receipt of the abstracts will be confirmed via e-mail.
- 4. The Abstracts will be presented as e-posters**

GUIDELINES FOR AUTHORS

The abstracts will be published as copy of the original text, so please follow the instructions below:

- Language: English.
- Abstract text word limit: 200 words (names and affiliation details are not included).
- Text alignment: Justified.
- Abstract title typed in: UPPER CASE.
- Authors full name (last name first) followed by initials and affiliation should follow the title of the abstract. The presenting author's name must be underlined.

The abstract should include the following:

Introduction, material and methods, results, conclusions.

ONLINE Abstract Submission
www.genetics2018.mdcongress.gr

ABSTRACTS DEADLINE
Tuesday, May 15, 2018

Abstract Submission

GENERAL INFORMATION

CONGRESS DATES

1-2 June, 2018

CONGRESS VENUE

Aegli Zappiou, Kipos Zappiou,
105 57 Athens Greece, T. 2103369300, F. 2103252952
Site:www.aeglizappiou.gr, E-mail: info@aeglizappiou.gr

Congress badges

Congress Badges are mandatory for the admission & access to the meeting halls and exhibition.

Internet availability

Wireless Internet access is available free of charge in the congress venue.

Language

The official language of the Congress is English. There will be no translation facilities available for the congress participants.

REGISTRATION FEES

Participants	70€
Trainees*	40€
Midwives/Nurses*	20€
Post graduate Students*	20€
Undergraduate Students*	Free

*Proof of status is required.

Participants and trainees registration fee includes:

- Access to scientific program
- Access to exhibition and poster area
- Full Congress material
- Certificate of attendance

Cancellation policy

(written request by email at md@mdcongress.gr or by fax at +302106074222)

- By May 1st, 2018, 50% of registration fee will be retained.
- After May 1st, 2018, 100% of registration fee will be retained.

ONLINE registrations
www.genetics2018.mdcongress.gr

Congress Organizing Bureau

MDcongress

118 Al. Panagouli str, 15343 Athens, Greece,
Tel.: +30.2106074200 - Fax: +30.210. 6074222
e-mail: md@mdcongress.gr, website: www.mdcongress.gr