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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](http://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*

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Fryssira Helena

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FRIDAY  
JUNE 1<sup>st</sup>  
2018

- 09:30-19.30 **Registrations**
- 10:00-10:30 **Welcome Addresses**  
**Introduction**  
*Helena Fryssira*
- 10:30-11:30 **1<sup>st</sup> Section: Metabolic and Mitochondrial disorders**  
*Chairs: Bartsocas C., Mastroyianni 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**
- 15:00-17:00 **2<sup>nd</sup> Section: Prenatal genetics**  
*Chairs: Sifakis S., Kolialexi A.*  
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.*  
Genetic Architecture of Human Disorders  
*Nicholas Katsanis*  
Rare Diseases: The use of new technologies in the diagnosis of congenital malformation syndromes and intellectual disability  
*Jane Hurst*
- 19:30 **WELCOME DRINK**

**SATURDAY**  
**JUNE 2<sup>nd</sup>**  
**2 0 1 8**

Scientific Program

- 09:30-17:00     **Registrations**
- 10:00-11:00     **3<sup>rd</sup> Section: Cancer Genomics**  
*Chairs: Tzetis M., Amenta S.*  
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     **4<sup>th</sup> Section: Neurodevelopmental disorders**  
*Chairs: Bacoula C., Fidani L.*  
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., 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](http://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](mailto: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](http://www.genetics2018.mdcongress.gr)

Congress Organizing Bureau

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