



"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 specialty 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

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FRIDAY
JUNE 1st
2 0 1 8

10:00-10:30 Welcome Addresses

Introduction Helena Fryssira

10:30-11:30 1st Section: Metabolic and Mitochondrial disorders

Chairs: Bartsocas C., Mastroyianni S.

Mitochondrial disorders *Graine Siobhan Gorman* 

Early diagnostic of Inborn Error 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



09:30-17:00	Registrations
10:00:11:00	3 <sup>rd</sup> Section: Cancer Genomics Chairs: Tzetis M., TBA 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

**ABSTRACTS DEADLINE** 

Tuesday, May 15, 2018

#### 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



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