

Bologna (Italy), 19th and 20th November, 2015



ALMA MATER STUDIORUM
UNIVERSITÀ DI BOLOGNA



SERVIZIO SANITARIO REGIONALE
EMILIA-ROMAGNA
Azienda Unità Sanitaria Locale di Bologna

International symposium on:

***THE IMPACT OF
GENOMIC DEEP SEQUENCING ON
PAEDIATRIC RESEARCH
AND CLINICAL PRACTICE***

Organized by
Paediatrics Unit
Sant'Orsola Academic Hospital
Alma Mater Studiorum
University of Bologna

Promoted by



FONDAZIONE
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MENARINI

**Stabat Mater Hall
Bologna, Italy**



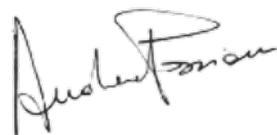
As many fields of medicine, paediatric research and clinical practice have greatly benefited, over this last decade, of the innovative discoveries derived by a deep approach to the genome.

We have selected, as icon for this symposium, the Angel holding a candlestick carved by a young Michelangelo for the shrine of San Domenico in Bologna, because the flame of genomic knowledge is like the candlestick lightening the dark scenario of our translational clinical practice.

The integration of the discoveries derived from deep genome sequencing has particular value for researchers who wish to address understudied issues in many different childhood diseases and clinical conditions.

The ultimate aim of this symposium is to promote a constructive dialogue between physicians and researches as to integrate distinctive knowledge in different fields towards a translational and personalized approach to paediatrics.

Andrea Pession
President of the symposium



Under the auspices of

Dipartimento Scienze Mediche e Chirurgiche
Alma Mater Studiorum
Università di Bologna

SIP
Società Italiana di Pediatria

CIRC
Centro Interdipartimentale di
ricerche sul cancro "Giorgio Prodi"
Alma Mater Studiorum
Università di Bologna

President of the symposium

Andrea Pession
Paediatrics Unit
Sant'Orsola Academic Hospital
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Organizing secretariat and CME provider

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03:00 p.m. Attendees registration

Welcome ceremony

03:30 p.m. Welcome addresses

F. Ubertini, Rector, University of Bologna

G. Corsello, President SIP- Società Italiana di Pediatria

A. Pession, President of the meeting

Opening lectures

Chairmen: G. Basso, A. Villani

04:00 p.m. **H. Hakonarson**

The future of precision medicine approaches in patient care

05:00 p.m. **S. Semplici**

Ethical issues

06:00 p.m. *Welcome cocktail*

Session I: Paediatric Immunology

Chairmen: G. Cocchi, C. Pignata

09:00 a.m. **C. Saunders**

Rapid whole-genome sequencing for genetic disease diagnosis in neonatal intensive care units

09:30 a.m. **A. Aiuti**

Novel approaches to screening and diagnosis of primary immunodeficiencies

Session II: Paediatric Rare Disease

Chairmen: L. Mazzanti, M. Seri

10:00 a.m. **S. Calvo**

Molecular diagnosis of infantile mitochondrial disease with targeted next-generation sequencing

10:30 a.m. **S. Vilarinho**

Individual exome analysis in diagnosis and management of paediatric liver diseases of indeterminate etiology

11:00 a.m. *Coffee break*

Session III: Paediatric Pneumology

Chairmen: F. Bernardi, S. Nava

11:30 a.m. **W.O. Cookson**

The GABRIEL experience and beyond: making sense of asthma with genes

12:00 a.m. **R.B. Parad**

Next generation sequencing of CFTR: utility in CF newborn screening

Session IV: Paediatric Endocrinology

Chairmen: A. Cassio, R. Pasquali

12:30 a.m. **C. Dionisi Vici**

Newborn screening

01:00 p.m. **P. Froguel**

Genetic characterization of monogenic childhood diabetes and obesity towards personalized metabolic medicine

01:30 p.m. *Lunch*

Session V: Paediatric Reumatology**Chairmen:** R. Meliconi, G. Ricci**03:00 p.m. A. Martini**
Genetics of auto-inflammatory disorders in childhood**03:30 p.m. G. Schulert**
Sequence data analysis with trios in rheumatic and other diseases**Session VI: Paediatric Gastroenterology****Chairmen:** G. Di Nardo, V. Stanghellini**04:00 p.m. S. Ennis**
Deep exome sequencing of paediatric autoimmune disease patients**04:30 p.m. P. Brigidi**
Functional and phylogenetic characterization of gut microbiome by deep sequencing**Session VII: Paediatric Oncology and Hematology****Chairmen:** F. Locatelli, G. Martinelli**05:00 p.m. C. Mullighan**
Genomic characterization of childhood acute lymphoblastic leukemia**05:30 p.m. R. Huether**
The landscape of somatic mutations in epigenetic regulators across 1,000 paediatric cancer genomes**06:00 p.m.** CME questionnaire**06:20 p.m.** Closing remarks

A. Aiuti	Milan, Italy
G. Basso	Padova, Italy
F. Bernardi	Bologna, Italy
P. Brigidi	Bologna, Italy
S. Calvo	Cambridge, USA
A. Cassio	Bologna, Italy
G. Cocchi	Bologna, Italy
W.O. Cookson	London, UK
G. Corsello	Palermo, Italy
G. Di Nardo	Rome, Italy
C. Dionisi Vici	Rome, Italy
S. Ennis	Southampton, UK
P. Froguel	London, UK
H. Hakonarson	Philadelphia, USA
R. Huether	Aliso Viejo, USA
F. Locatelli	Rome, Italy
G. Martinelli	Bologna, Italy
A. Martini	Genova, Italy
L. Mazzanti	Bologna, Italy
R. Meliconi	Bologna, Italy
C.G. Mullighan	Memphis, USA
S. Nava	Bologna, Italy
R.B. Parad	Boston, USA
R. Pasquali	Bologna, Italy
A. Pession	Bologna, Italy
C. Pignata	Naples, Italy
G. Ricci	Bologna, Italy
C. Saunders	Kansas City, USA
G. Schulert	Cincinnati, USA
S. Semplici	Rome, Italy
M. Seri	Bologna, Italy
V. Stanghellini	Bologna, Italy
S. Vilarinho	New Haven, USA
A. Villani	Rome, Italy

GENERAL INFORMATION

Meeting venue

The venue for the symposium is
Stabat Mater Hall
Archiginnasio Public Library
Piazza Galvani 1, Bologna (Italy)

Secretariat during the meeting

The Secretariat will be open at the following times:
Thursday 19th November from 03:00 p.m. to 06:00 p.m.
Friday 20th November from 09:00 a.m. to 06:30 p.m.

Official language

The official language of the meeting will be English.
Simultaneous translation will not be provided.

CME: Continuing Medical Education

DueCi Promotion srl, CME Provider of the symposium, will assign CME credits to the following profiles:

- **Physicians** specialized in:
 - Paediatrics
 - Hematology
 - Endocrinology
 - Oncology
 - Gastroenterology
 - Medical genetics
 - Metabolic disease and diabetology
 - Internal medicine
 - Rheumatology
- **Biologists**

Attendees' registration

The symposium is free to attend, but on line registration is required.
Please link to the organizing secretariat website:
www.duecipromotion.com > Congressi Medici
> Calendario Congressi > Registrazione on line

On line registrations will be accepted on a first-come, first-served basis and may be closed in advance whenever the maximum number of participants allowed in the venue will be reached.

Registration includes:

access to scientific sessions, welcome cocktail, coffee breaks, lunch, program book, certificate of attendance.



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