

Bologna (Italy), 19<sup>th</sup> and 20<sup>th</sup> 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



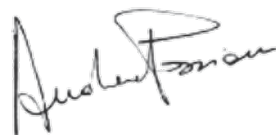
As many fields of medicine, paediatric research and clinical practice have greatly benefited, over this last decade, of the innovative discoveries derived by a deep approach to the genome.

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 researchers 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  
Alma Mater Studiorum  
Università di Bologna

### Scientific secretariat

**Annalisa Astolfi**  
**Riccardo Masetti**

### Promoted by

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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. Corsello, G. Basso

**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:** M. Duse, G. Faldella

**09:00 a.m.** **S.F. Kingsmore**  
 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 Reumathology****Chairmen:** R. Meliconi, G. Ricci**03:00 p.m. A. Martini**  
Genetics of auto-inflammatory disorders in childhood**03:30 p.m. J.B. Harley**  
Sequence data analysis with trios in rheumatic and other diseases**Session VI: Paediatric Gastroenterology****Chairmen:** S. Cucchiara, 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

<b>A. Aiuti</b>	Milan, Italy
<b>G. Basso</b>	Padova, Italy
<b>F. Bernardi</b>	Bologna, Italy
<b>P. Brigidi</b>	Bologna, Italy
<b>S. Calvo</b>	Cambridge, USA
<b>A. Cassio</b>	Bologna, Italy
<b>W.O. Cookson</b>	London, UK
<b>G. Corsello</b>	Palermo, Italy
<b>S. Cucchiara</b>	Rome, Italy
<b>C. Dionisi Vici</b>	Rome, Italy
<b>M. Duse</b>	Rome, Italy
<b>S. Ennis</b>	Southampton, UK
<b>G. Faldella</b>	Bologna, Italy
<b>P. Froguel</b>	London, UK
<b>H. Hakonarson</b>	Philadelphia, USA
<b>J.B. Harley</b>	Cincinnati, USA
<b>R. Huether</b>	Aliso Viejo, USA
<b>S.F. Kingsmore</b>	Kansas City, USA
<b>F. Locatelli</b>	Rome, Italy
<b>G. Martinelli</b>	Bologna, Italy
<b>A. Martini</b>	Genova, Italy
<b>L. Mazzanti</b>	Bologna, Italy
<b>R. Meliconi</b>	Bologna, Italy
<b>C.G. Mullighan</b>	Memphis, USA
<b>S. Nava</b>	Bologna, Italy
<b>R.B. Parad</b>	Boston, USA
<b>R. Pasquali</b>	Bologna, Italy
<b>A. Pession</b>	Bologna, Italy
<b>G. Ricci</b>	Bologna, Italy
<b>S. Semplici</b>	Rome, Italy
<b>M. Seri</b>	Bologna, Italy
<b>V. Stanghellini</b>	Bologna, Italy
<b>S. Vilarinho</b>	New Haven, USA

# 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 19<sup>th</sup> November from 03:00 p.m. to 06:00 p.m.  
Friday 20<sup>th</sup> 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](http://www.duecipromotion.com) > Congressi Medici  
> Calendario Congressi > Registrazione on line

## Registration deadline: 25<sup>th</sup> October, 2015

**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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