CONFERENCE

15th Conference of Undiagnosed Diseases Network International

16-17 October 2026

Università di Siena
Crypt Hall - San Francesco Campus
Siena, Italy

Coordinator:
Professor Alessandra Renieri

Scientific Committee:
William A. Gahl
Roberto Giugliani
Ratna Dua Puri
Alessandra Renieri

un evento organizzato da | BioMedia La condivisione del sapere

15th CONFERENCE OF UNDIAGNOSED DISEASE NETWORK INTERNATIONAL

SIENA, OCTOBER 16–17, 2026

Endorsements

Azienda ospedaliero-universitaria Senese

COMUNE DI SIENA

con il patrocinio del Comune di Siena

ERKNet
The European
Rare Kidney Disease
Reference Network

European Reference Network | Genetic Tumour Risk Syndromes (ERN GENTURIS)

ITHACA

SIGU
Società Italiana di Genetica Umana

UNIVERSITÀ DI SIENA
1240

15th CONFERENCE OF UNDIAGNOSED DISEASE NETWORK INTERNATIONAL

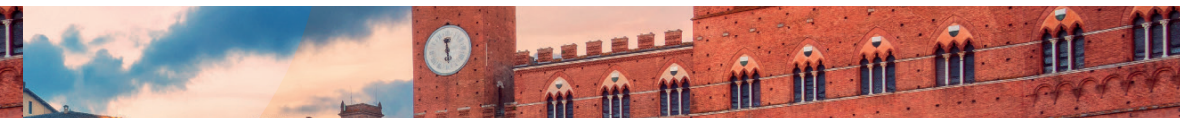
SIENA - OCTOBER 16–17, 2026

FACULTY:

Ozlem Akgun-Dogan (Turkey)
Yasemin Alanay (Turkey)
Hind Alsharhan (Kuwait)
Francesca Ariani (Italy)
Maria Barrero (Spain)
Claudia Basagni (Italy)
Mario Capasso (Italy)
Laura Cardarelli (Italy)
Helene Cederroth (Sweden)
Andra Ciucă (Romania)
Fabio Coppedè (Italy)
Domenico Coviello (Italy)
Nicola Dimitri (Italy)
William A. Gahl (USA)
Livia Garavelli (Italy)
Mattia Gentile (Italy)
Emiliano Giardina (Italy)
Elisa Giorgio (Italy)
Roberto Giugliani (Brazil)
Stefano Gustincich (Italy)
Reham Khalaf-Nazzal (UK)
Thomas Liehr (Germany)
Giuseppe Matullo (Italy)
Béla Melegh (Hungary)
Giuseppe Merla (Italy)
Ilaria Meloni (Italy)
Manuela Morleo (Italy)
Vincenzo Nigro (Italy)
Uğur Özbek (Turkey)
Tayfun Ozcelik (Turkey)
Stephen C. Pak (USA)
Federico Perino (Italy)
Angela Peron (Italy)
Graziano Pesole (Italy)
Anna Maria Pinto (Italy)
Ratna Dua Puri (India)
Clementina Radio (Italy)
Ilona Reischl-Kok (Austria)
Alessandra Renieri (Italy)
Moin Saleem (UK)
Marco Salvatore (Italy)
Nara Sobreira (USA)
Rita Selvatici (Italy)
Marco Tartaglia (Italy)
Domenica Taruscio (Italy)
Tinatin Tkemaladze (Georgia)
Wendy A.G. Van Zelst-Stams (Netherlands)

EVENT VENUE

University of Siena – Crypt Hall – san Francesco Campus
Piazza San Francesco 7, Siena



FRIDAY, OCTOBER 16, 2026

08:00 SESSION 1

OPENING SESSION
Co-chair: ***Livia Garavelli (Italy)***
Co-chair: ***Ilaria Meloni (Italy)***

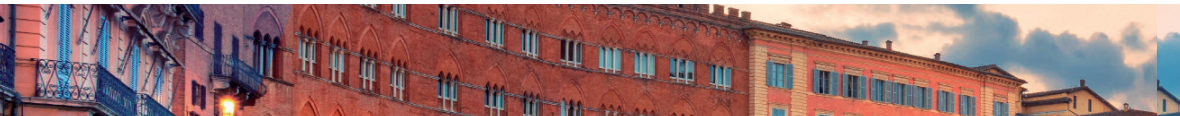
Welcome to the 15th UDNI
Prof. Alessandra Renieri (Siena)
Dr. Ettore Ruggi d'Aragona, Coordinator of the National Committee for Rare Diseases (CoNaMaR)
Prof. Donata Medaglini Vice-Rector of University of Siena
Dr. Claudia Basagni, Rare Disorder Coorinator AOU Senese
Dr. Livia Garavelli (Reggio Emilia) SIGU
The origins of UDNI and lessons learned through the years
William Gahl (USA)

From UDPs to Hackathons
Helene Cederroth (Sweden)

09:00-10:00 SESSION 2

PANEL: SYSTEMATIC GENOMIC REANALYSIS AND GENETIC COUNSELLING
Co-chair: ***Yasemin Alanay (Turkey)***
Co-chair: ***Clementina Radio (Italy)***

Systematic genomic reanalysis as a scalable strategy in undiagnosed disease programs
Ozlem Akgun-Dogan (Turkey)



The Role of EBMG in undiagnosed diseases
Angela Peron (Italy)

The Role of Genetic Counselling in Rare and Undiagnosed Diseases
Andra Ciucă (Romania) on line

How European Board of Medical Genetics serves the needs of patients who use genetic services in Europe
Thomas Liehr (Germany) on line

10:00-10:30 **Break – Poster Viewing1 (CHIOSTRO)**

10:30-13:30 **SESSION 3**

PANEL: NEW STRATEGIES AND ADVANCED TECHNOLOGIES FOR INCREASING DIAGNOSTIC YELD
Co-chair: ***Marco Tartaglia (Italy)***
Co-chair: ***Fabio Coppedè (Italy)***

Evaluating DNA methylation episignatures as a first-tier diagnostic test in individuals with suspected genetic disorders
Tinatin Tkemaladze (Georgia)

The "Undiagnosed Patients Program" at Ospedale Pediatrico Bambino Gesù: novel disease gene discovery and diagnostic impact
Marco Tartaglia (Italy)

Clinical and epigenetic characterisation of a novel Kleefstra-like syndrome due to EHMT2 variants
Maria Barrero (Spain)

Integrative and systematic genomic approaches to improve diagnosis in rare and undiagnosed diseases
Uğur Özbek (Turkey)

LR-WGS: one fit all analyses
Alessandra Renieri (Italy)

Novel disease gene discovery within the Telethon Undiagnosed Diseases Program
Manuela Morleo – Telethon (Italy)

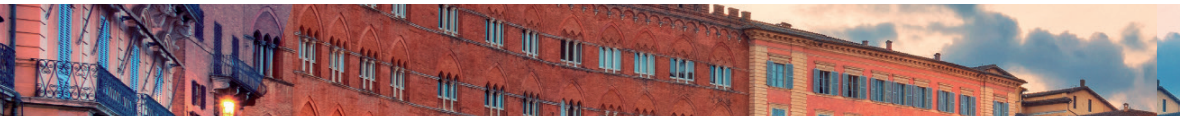
Sharing FAIR data and 1 Million Genomes European initiative
Domenico Coviello (Italy)

ORAL PRESENTATIONS OF SELECTED ABSTRACTS

- DNA methylation profiling of undiagnosed patients: a five-year experience with orthogonal omics approach to increase diagnostic yield in rare diseases – ***(Andrea Ciolfi)***

- Implementation of an integrated telemedicine and multi-omics workflow to improve diagnostic yield in undiagnosed patients: the experience of RETE IDEA, the Italian Network of Pediatric IRCCS - ***(Francesca Clementina Radio)***

13:30-14.00 **Lunch**



15:00-17:30 SESSION 5

PANEL: DATA SHARING AND FUNCTIONAL ANALYSIS TO FACILITATE GENE DISCOVERY, VARIANT CLASSIFICATION AND PATIENT DIAGNOSIS
Co-chair: ***Francesca Ariani (Italy)***
Co-chair: ***Wendy A.G. van Zelst-Stams (Nijmegen)***

Accelerating rare disease diagnosis using C. elegans
Stephen C. Pak (USA)

International data sharing
Nara Sobreira (USA)

The Network of Italian Genomes NIG as a Reference Framework for Rare Diseases and Genomics Initiatives in Italy
Giuseppe Matullo (Italy)

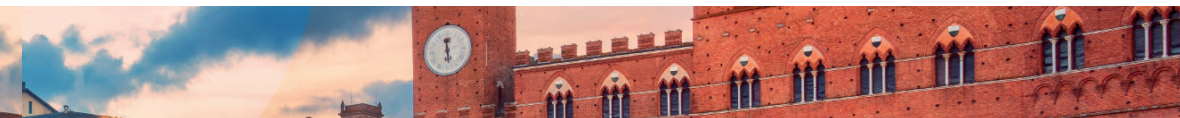
The Italian approach to rare diseases: the GENERA project
Vincenzo Nigro (Italy)

UDP Programs in Italy: Building the Future for Patients Without a Diagnosis
Marco Salvatore (Italy)

ELIXIR-IT: The Italian Research Infrastructure for Big Data in Life Sciences
Graziano Pesole (Italy)

The ERDERA Project
Holm Graessner (Germany) on-line

17:00-17:30 Break – Poster Viewing (CHIOSTRO)



17:30-18:30 SESSION 6

PANEL: NEWBORN SCREENING DISEASES
Co-chair: ***Laura Cardarelli (Italy)***
Co-chair: ***Tinatin Tkemaladze (Georgia)***

The Challenge of Genetic and Genomic Newborn Screening: Screen4Care and GenNewBs projects
Rita Selvatici (Italy)

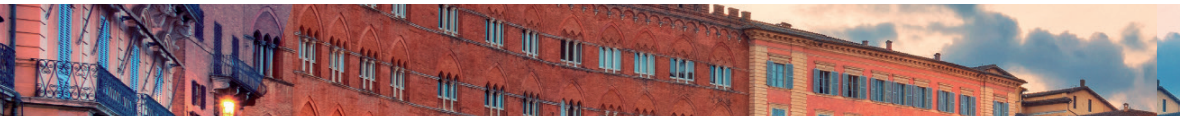
From the Pilot Project to the Public Health Implementation: gNBS as an Opportunity for All Newborns.
Genoma Puglia Program, Italy
Mattia Gentile (Italy)

18:30-19:30 SESSION 7

ORAL PRESENTATIONS OF SELECTED ABSTRACTS
Co-chair: ***Mario Capasso (Italy)***
Co-chair: ***Emiliano Giardina (Italy)***

- whole genome sequencing identifies a 20.4 kb homozygous deletion in dnajb4, providing a diagnosis that can radically alter clinical management – ***(Ugur Ozbek)***

- An unsolved case of intellectual disability, autism, and epilepsy associated with a de novo variant in prrc2a, a novel m6a mrna methylation reader – ***(Bryn Webb)***



- context-aware automated hpo annotation of russian clinical texts using language and graph models – ***(Artem Borovikov)***

- Systematic Detection of Undiagnosed Monogenic Kidney Disease in Routine Adult Nephrology Care – A Real-World Experience ***(Roberta Fenoglio)***

- From Global Model to Internal Engine: Lessons from the 2025 Undiagnosed Hackathon and the Design of a Dual-Track Successor at Mayo Clinic – ***(Cherisse Marcou)***

- DNA methylation profiling via long read sequencing in patients with SETD1B-related disorder – ***(Virdo' Simona)***

SATURDAY, OCTOBER 17, 2026

08:30-10:15 SESSION 8

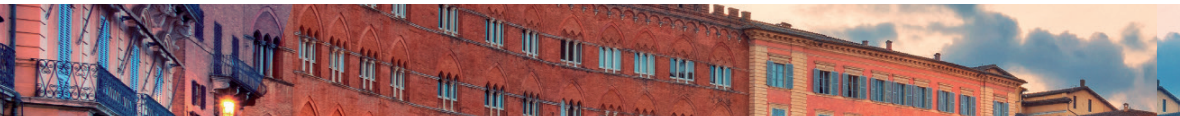
PANEL: NETWORKS, EDUCATION AND EMERGING REGIONS ON UNDIAGNOSED DISEASES
Co-chair: ***Anna Maria Pinto (Italy)***
Co-chair: ***Ratna Dua Puri (India)***

Building and expanding undiagnosed disease programs in emerging regions
Hind Alsharhan (Kuwait)

REBRAND - The Brazilian Network of Undiagnosed Diseases
Roberto Giugliani (Brazil)

UDNI Low and Middle Income Countries (LMICs) Working Group: Aims, Structure and Results
Domenica Taruscio (Italy)

Proficiency Testing and Certification
Béla Melegh (Hungary)



10:15-11:15 **ORAL PRESENTATIONS OF SELECTED ABSTRACTS**

Co-chair: ***Federica Perino (Italy)***
Co-chair: ***Maria Chiara Siciliano (Italy)***

-Whole genome reanalysis improves diagnostic yield in frequent SORD-associated hereditary motor neuropathy – ***(Murtazina Aysylu)***

-A second independent family with biallelic lrp1 variants provides further evidence for a candidate autosomal recessive lrp1-related disorder ***(Ayca Yigit)***

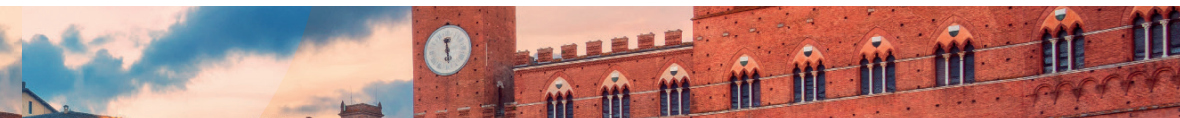
-Candidate Modifier Gene Variants and Phenotypic Variability in Georgian Patients with Cystic Fibrosis ***(Eka Kvaratskhelia)***

- Accelerating genetic diagnosis in the NICU via machine learning assisted selection of infants for genome sequencing ***(Stephen Meyn)***

- Resolving a 10-Year Diagnostic Odyssey: Identification of a Rare Pathogenic PQBP1 Variant in a 12-Year-Old Boy with an Undiagnosed Neurodevelopmental Disorder ***(Luigia De Falco)***

- From global to local ancestry: refining rare disease variant interpretation in admixed individuals ***(Béla Melegh)***

11: 15-11:45 **Break – Poster Viewing (CHIOSTRO)**



11:45-13:00 SESSION 9

PANEL: FROM DIAGNOSES TO THERAPIES
Co-chair: ***Alessandra Renieri (Italy)***
Co-chair: ***Nicola Dimitri (Italy)***

The role of CAT in Advanced Therapy Medicinal Products ATMPs
Ilona Reischl-Kok chair of CAT EMA – on-line (Austria)

Gene therapy in nephrotic syndromes
Moin Saleem (UK)

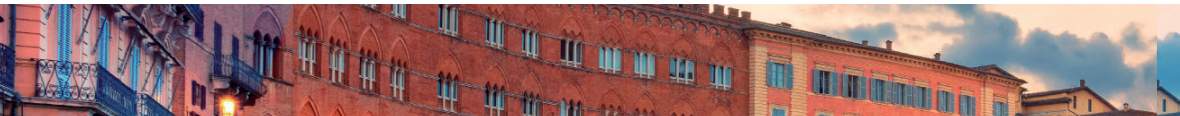
14:00-15:00 Lunch

15:00-16:00 CLOSING SESSION
Co-chair: ***William Gahl (USA)***
Co-chair: ***Helene Cederroth (Sweden)***

LECTIO MAGISTRALIS Burden of inherited predisposition to genetic diseases in diverse populations
Tayfun Ozcelik (Turkey)

Abstract Prize
Introduction to Next UDNI Conference

Closing
William Gahl (USA), Helene Cederroth (Sweden) and Alessandra Renieri (Italy)



POSTER

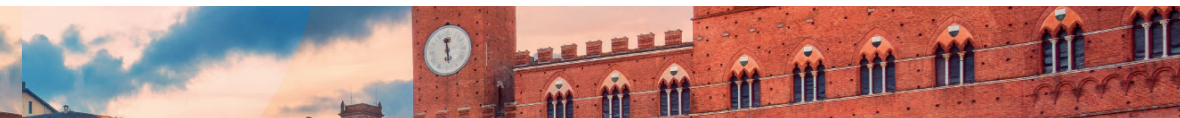
16:00-18:30 **UDNI BUSINESS AND COMMITTEE MEETINGS**
(open to UDNI members and non-members)
William Gahl (USA)

P01 – A De Novo Heterozygous Loss-of-Function ASPM Variant in a Child with Autism Spectrum Disorder Beyond an Established Metabolic Diagnosis
Topaloglu Mehmet Mert, Yigit Ayca, Gunes Seda, Ozbek Ugur

P02 – A solved case of imagawa-matsumoto syndrome with a de novo intronic suz12 variant and novel findings
Margolin Lia, Webb Bryn, Keppler-Noreuil Kim

P03 – Abnormal pigmentation, premature graying of hair, nail and dental anomalies and neurological findings in two siblings with Cobalamin J deficiency
Rock Rachel, Raved Dani, Anikster Yair

P04 – Economic analysis of the care pathway in Rare Diseases: Evidence from a specialized center in Brazil
Scherer Luciene C., Barbosa Fabrizio, Fischinger Moura de Souza Carolina, Riegel-Giugliani Mariluce, Oliveira Bibiana Mello de, Giugliani Roberto

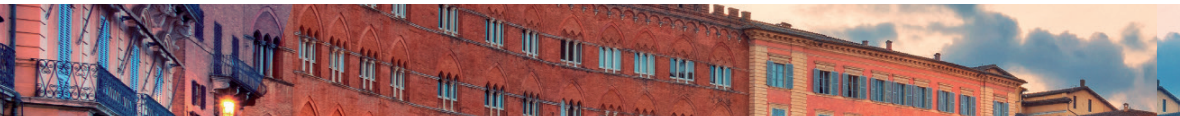


P05 – Integrating Optical Genome Mapping and DNA Methylation Profiling for Rapid Molecular Diagnosis of Facioscapulohumeral Muscular Dystrophy
Megalizzi Domenica, Strafella Claudia, Trastulli Giulia, Proietti Piorgo Emma, Papasergi Bruno, Colantoni Luca, Peconi Cristina, Cascella Raffaella, Buoncuore Giorgia, Torreri Paola, Salvucci Benedetta, Caltagirone Carlo, Salvatore Marco, Giardina Emiliano

P06 – Patterns of Undiagnosed Diseases over 18 Years
Adams David, Gahl William, Tifft Cynthia, Toro Camilo, Wolfe Lynne, D'Souza Precilla, Maduro Valerie, Malicdan May, Macnamara Ellen, Acosta Maria, Novacic Danica

P07 – Rare variants disrupting mRNA cleavage and polyadenylation associated with undiagnosed rare disorders
Kainov Yaroslav, Dias Cristina, Hubbard Tim

P08 – Single exome sequencing for non-critically ill children hospitalized in the general pediatric ward: Diagnostic yield, lessons and challenges
Chorin Odelia, Barel Ortal, Lev-Hochberg Shelly, Ityel Hadas, Akselrod Dikla, Rein-Rothschild Annick, Efargan Adi, Vivante Asaf, Somech Raz, Semo-Oz Rotem, Bezalel Yael, Eliyahu Aviva, Regev Miriam, Greenbaum Lior, Pinchevski-Kadir Shiran, Heimer Gali, Arkush Leo, Bolkier Yoav, Pessach Itai M., Birk Ohad S., Dominissini Dan, Pode-Shakked Ben



P09 – Supporting the Rare Disease Ghana Initiative: Objectives, Key Results, Challenges, and Lessons Learned in Providing Diagnoses for Undiagnosed Rare Diseases in Ghana

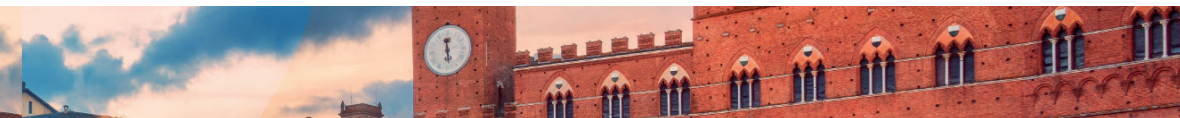
Marchionni Enrica, D'Apice Maria Rosaria, Terracciano Alessandra, Daga Sergio, Pisaneschi Elisa, Sangiuolo Federica, Novelli Antonio, Wiafe Samuel Agyei, Novelli Giuseppe

P10 – Impaired lymphatic drainage in a child with hydrops fetalis and a novel variant in the PROX1 gene: case report and review of literature

Pisaneschi Elisa, De Rose Domenico Umberto, Pugnaloni Flaminia, Costa Simonetta, Menzella Nicoletta, Diociaiuti Andrea, Natali Gian Luigi, Secinaro Aurelio, Massella Laura, Digilio Maria Cristina, Campi Francesca, Novelli Antonio, Conforti Andrea, Capolupo Irma, Vento Giovanni, Dotta Andrea

P11 – When the diagnosis doesn't fit: dna methylation signatures that revise the diagnosis

Ityel Hadas, Dilian Yael, Glick-Saar Efrat, Mor Nofar, Heimer Gali, Cohen Louisa, Chorin Odelia, Lev-Hochberg Shelly, Akselrod Dikla, Efargan Adi, Eliyahu Aviva, Regev Miriam, Rein-Rothschild Annick, Birk Ohad S, Dominissini Dan, Pode-Shakked Ben

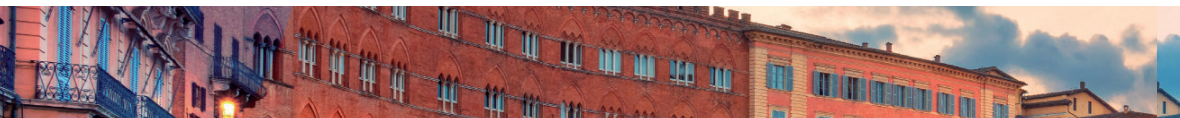


P12 – Diagnostic challenges in hereditary ovarian cancer: discordant somatic and germline findings in a rad51d-associated case-
Rosu Cristian-Matei, Antonescu Madalina-Doina, Vasiliniuc Anisia-Cristiana, Buica Vlad, Russell Lindsay, Perdun Patricia, Hotinceanu Iulian-Andrei, Pop Corina-Silvia, Craiu Dana-Cristina, Bucur Octavian, Nicoli Elena-Raluca

P13 – A tiered functional evidence framework for rare disease diagnosis in the nih undiagnosed diseases program
Malicdan May Christine, Adams David, Macnamara Ellen, Toro Camilo, Wolfe Lynne, Ahmed Asiya, D'Souza Precilla, Maduro Valerie, Novacic Danica, Tifft Cynthia, Gahl William

P14 – Diagnostic challenges in an undiagnosed odyssey: a case report on a kcp gene variant of uncertain significance
Vasiliniuc Anisia-Cristiana, Antonescu Madalina-Doina, Buica Vlad, Russell Lindsay, Perdun Patricia, Rosu Cristian-Matei, Jurcut Ruxandra Oana, Jurcut Ciprian-Viorel, Pop Corina-Silvia, Craiu Dana-Cristina, Bucur Octavian, Nicoli Elena-Raluca

P15 – ONT methylation analysis contributes to explain a discordant phenotype in sisters with Rett syndrome
Pesaresi Simone, Serena Deborah, Dahine A, Adduci Mariarosaria, Ariani Francesca, Fallerini Chiara, Narducci Chiara,Scandurra Valeria, Buoni Sabrina, Meloni Ilaria, Furini Simone, Renieri Alessandra



P16 – Exome analysis on cfDNA detects post-zygotic event in undiagnosed patients with neurodevelopmental/autism spectrum disorders
Miriana Danelli, Mariarosaria Adduci, Giorgio Signori, Caterina Lo Rizzo, Anna Maria Pinto, Simone Pesaresi, Camilla Ruta, Simone Furini, Alessandra Renieri, Chiara Fallerini

P17 – Molecular optimization of preimplantation genetic testing using a high-resolution CNV detection platform.
Giudice Alessandra, Russo Giulia, Bruttini Mirella, Ricci Sandra, Tosti Claudia, Ciociola Francesca, Lo Rizzo Caterina, Renieri Alessandra

P18 – ctDNA NGS detects clinically relevant genomic targets in head and neck cancer
Mariarosaria Adduci, Giorgio Signori, Caterina Lo Rizzo, Anna Maria Pinto, Miriana Danelli, Camilla Ruta, Maria Chiara Siciliano. Cristiana Bellan, Ignazio Martellucci, Chiara Fallerini, Marco Mandala, Alessandra Renieri

P19 – Biallelic TET2 variants of clonal haematopoiesis in VEXAS- like patients
Siciliano Maria Chiara, Vitale Antonio, Dahine Antoine, Lo Rizzo Caterina, Leoncini Lorenzo, Cantarini Luca, Lazzi Stefano Renieri Alessandra

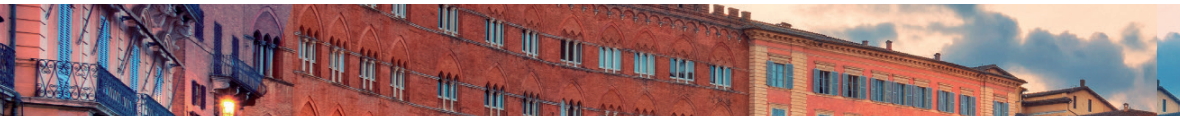
SIMPOSI

FRIDAY, OCTOBER 16, 2026

14:00-15:00 **Satellite Symposia (30 min each): NO ECM**
Sponsored by Company 1: 4bases Italia
Sponsored by Company 2: Oxford Nanopore Technologies plc

SATURDAY, OCTOBER 17, 2026

13:00-14:00 **Satellite Symposia (30 min each): NO ECM**
Sponsored by Company 3: Illumina
Sponsored by Company 4: enGenome



GENERAL INFORMATION

Free Registration

Registration is mandatory via the following website:
https://biomedia.net/17294/

Registration Procedure

The online registration form can be completed directly on the website www.biomedia.net, on the page dedicated to the event.

Ecm Credits

ECM credits and the certificate of attendance will be awarded only to participants attending the event in person in Siena.The course is accredited by Biomedia, Provider No. 148, under the Italian Ministry of Health's National Continuing Medical Education Programme (ECM), for the following professional categories: **Categories**: Biologist; Medical Doctor; Biomedical Laboratory Technician.

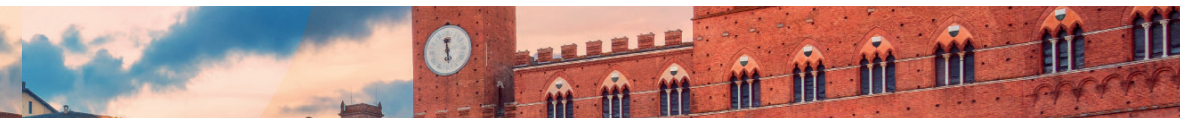
Disciplines: Cardiology; General Surgery; Pediatric Surgery; Dermatology and Venereology; Hematology; Endocrinology; Medical Genetics; Respiratory Medicine; Endocrinology and Diabetology (Metabolic Diseases); Nephrology; Neonatology; Neurology; Child Neuropsychiatry; Ophthalmology; Oncology; Pediatrics; Rheumatology.

To be eligible for ECM (Continuing Medical Education) credits:

To be eligible for ECM credits, participants must:

- attend at least 90% of the educational activities;
- sign the attendance register at the beginning and end of the day;
- complete and submit the satisfaction, evaluation and learning questionnaire available at:

https://elearning.biomedia.net/gestione/



The ECM test will be available online for 3 days, starting from the day after the event.Once the questionnaire has been successfully completed, participants will be able to download their ECM certificate directly from the website.In accordance with the AGENAS Notice of 23 June 2014, please note that the online assessment test may be completed only once and cannot be repeated.

For further information:

http://ape.agenas.it/

Participants are reminded that, for the 2026–2028 three-year reference period, the maximum number of training credits that may be obtained through participation upon invitation by a sponsor is limited to one third of the total credits required.

CERTIFICATE OF ATTENDANCE

All duly registered participants attending the event in person in Siena will be entitled to a certificate of attendance.The certificate will not be issued in paper format. Participants will be able to download and print it from the day the event ends via the following platform:

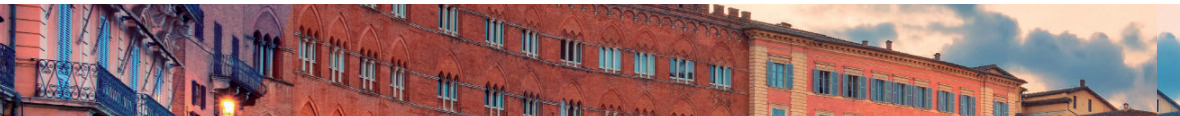
https://elearning.biomedia.net

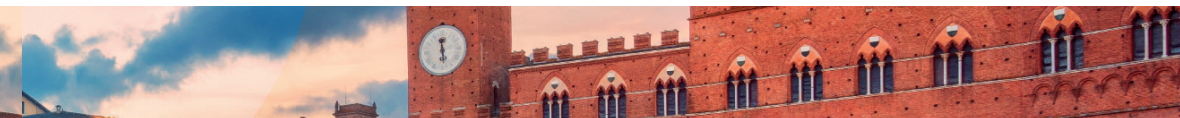
If you have lost your login credentials, please request new access credentials through the event area at:

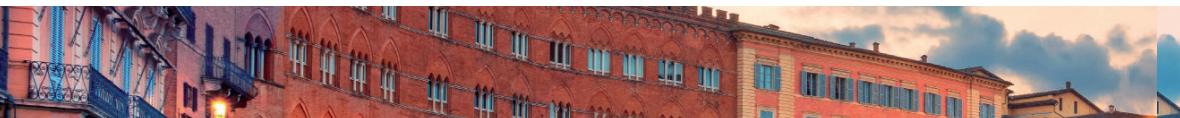
https://elearning.biomedia.net

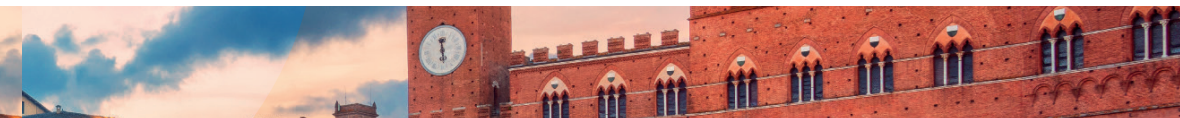
COURSE VENUE

Meeting venue:
Aula Cripta - Complesso San Francesco Università di Siena -Siena Italy









with the unconditional contribution of

enGenome

4bases

illumina®

Philogen
innovating targeting

Oxford Nanopore Technologies

TECHNO GENETICS
KHB GROUP

TEMAVita
Amici per Sempre
Mutua con Socio Sostenitore Banca TEMA

CeGaT

un evento organizzato da BioMedia
La condivisione del sapere

E-mail: convegni@biomedia.net
Sito: www.biomedia.net