



# HUMAN GENOME MEETING 2024

8-10 April 2024, Rome - Italy



# SCIENTIFIC PROGRAMME

Under the patronage of





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# Patronages

Under the patronage of



# Organizing committee

## **Giuseppe Novelli**

*Chairman*

Professor of Medical Genetics, Tor Vergata University of Rome, Italy  
Adjunct Professor, Department of Pharmacology, University of Nevada, Reno, NV, USA

## **Juergen Reichardt**

*Co-chair and Hugo liaison*

Adjunct Professor, Australian Institute of Tropical Health and Medicine, James Cook University, Australia

## **Paola Grammatico**

Professor of Medical Genetics, Sapienza University of Rome, Italy

## **Piero Carninci**

Deputy Director, Riken Center for Integrative Medical Sciences, Riken Yokohama Campus, Yokohama, Japan  
Human Technopole, Milan, Italy

# Scientific committee

## **Karen Avraham**

Tel Aviv University Israel

## **Piero Carninci**

Human Technopole Italy

## **Ada Hamosh**

Johns Hopkins University US

## **Giuseppe Novelli**

Tor Vergata University of Rome Italy and  
University of Nevada USA

## **Mike Snyder**

Stanford University US

## **Anne Bowcock**

Mount Sinai US

## **Giorgia Giotto**

University of Trieste Italy

## **Partha Majumder**

National Institute of Biomedical  
Genomics India

## **Juergen Reichardt**

James Cook University Australia

## **Birutė Tumienė**

Vilnius University Hospital Santaros Klinikos  
Lithuania



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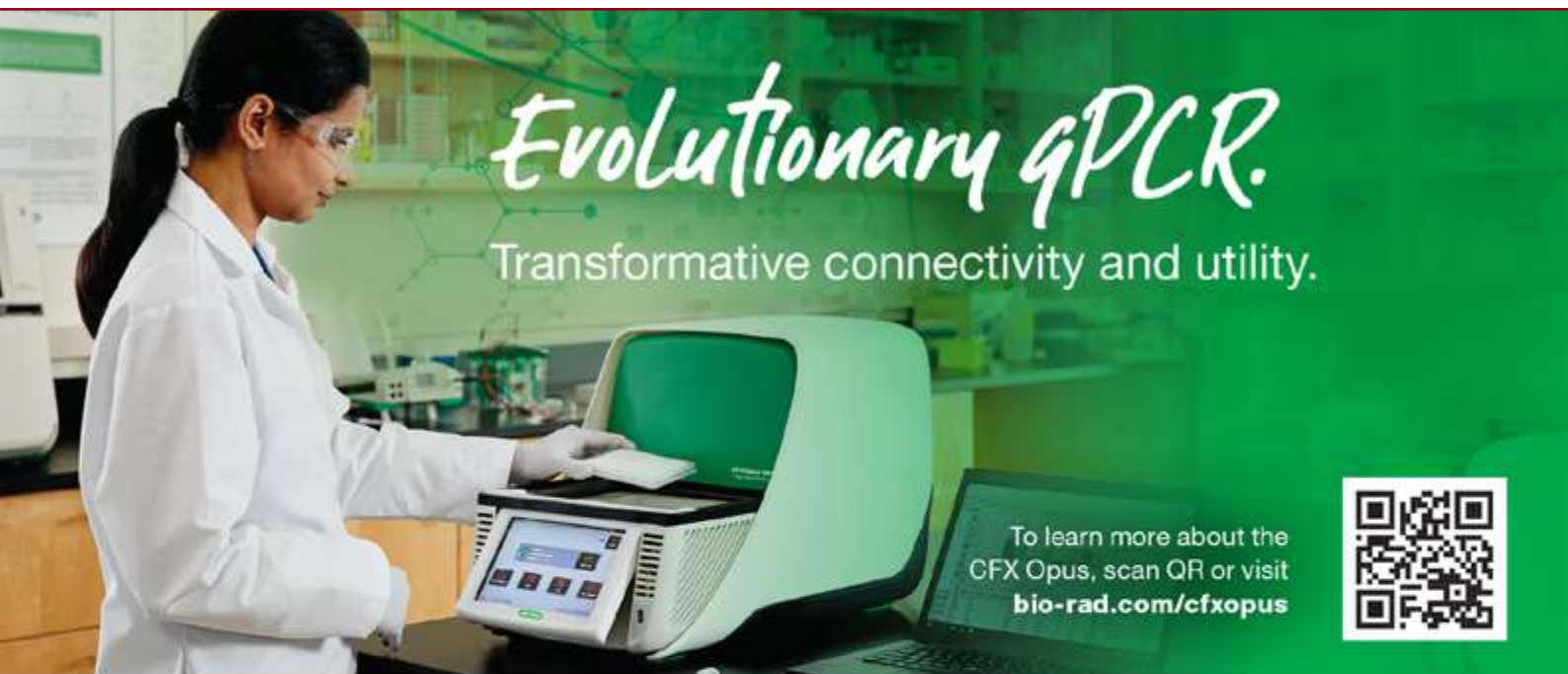
 When Precision Matters

For over 10 years, 4bases' mission is to address increasing medical needs in the fields of prevention and precision medicine. As a Swiss-based company with a production facility in Italy, 4bases is specialized in the development, production, and commercialization of clinically validated reagent (CE-IVD) kits for NGS for diagnostic purposes. Our kits are compatible with Illumina, ThermoFisher, MGI and Oxford Nanopore Technologies sequencers. On the data analysis side as well, they can be used either in combination with the most advanced commercial solutions or with 4eVAR, our proprietary software. Our offer covers a wide range of diagnostic solutions from single gene panels to whole exome or clinical exome analysis, including short and long reads solutions and a high degree of versatility that will accommodate your specific needs. Let's meet on booth #17-18 to discuss them!

---


**Meet Our Team!**

At the Human Genome Meeting 2024, our Team of experts will hold a workshop: connect with us and learn more on how sequencing can impact precision medicine in a clinical context! We'll wait for you on Tuesday, April 9 at 2 pm, in the second plenary.

*Evolutionary qPCR.*  
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### Introducing the new CFX Opus Real-Time PCR Systems

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# Human Genomics is a peer-reviewed, open access, online journal

that focuses on the application of genomic analysis in all aspects of human health and disease, as well as genomic analysis of drug efficacy and safety, and comparative genomics.

Topics covered by the journal include, but are not limited to: pharmacogenomics, genome-wide association studies, genome-wide sequencing, exome sequencing, next-generation deep-sequencing, functional genomics, epigenomics, translational genomics, expression profiling, proteomics, bioinformatics, animal models, statistical genetics, genetic epidemiology, human population genetics and comparative genomics.

## 2022 Citation Impact

4.5 - 2-year Impact Factor

4.9 - 5-year Impact Factor

## 2022 Usage

510,313 downloads

1,940 Altmetric mentions

## Editorial Board

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## Why publish with us?

- Focus on the application of genomic analysis in all aspects of human health and disease
- Official journal of the Human Genome Organization
- Maximum visibility with open access

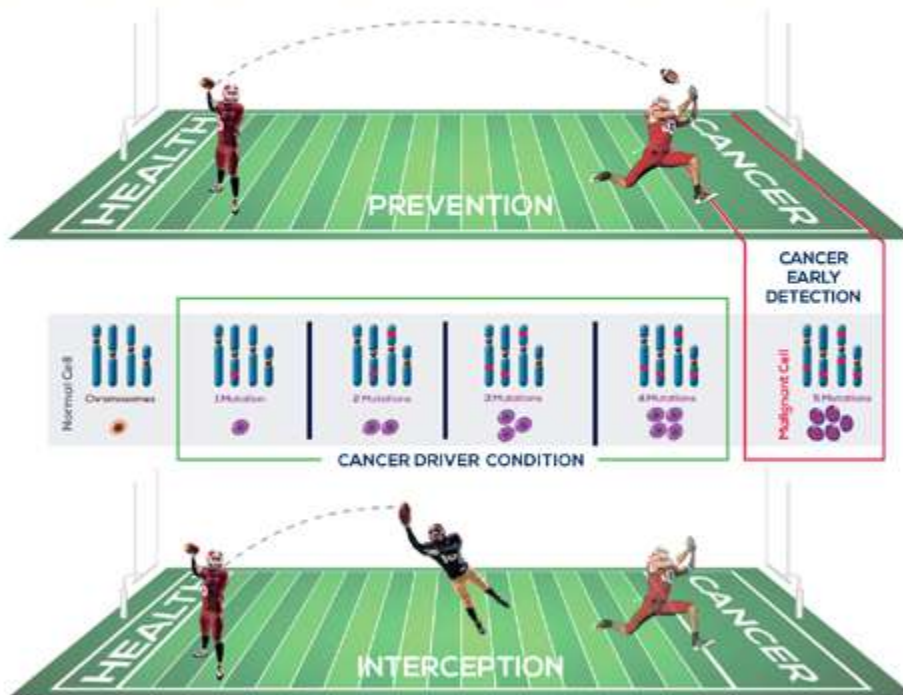
Submit your manuscript at: [humgenomics.biomedcentral.com/about](https://humgenomics.biomedcentral.com/about)

and see our Collections on key topics in the field (e.g., gene family updates, pharmacogenomics, urban wastewater genome analysis to track infectious disease) at: [humgenomics.biomedcentral.com/articles/collections](https://humgenomics.biomedcentral.com/articles/collections)

\* The Impact Factor (IF) is just one measure used for evaluating the impact of research. For further information on the IF and alternative metrics used to measure the impact of research, please visit [biomedcentral.com/p/bmc-impact](https://biomedcentral.com/p/bmc-impact)

# CANCER DRIVER INTERCEPTION

Today it is possible to switch the focus from early cancer detection and generic external risk factors reduction to actionable cancer driver interception.



Adapted from: Blackburn EH. Cancer Prev Res (Phila). 2011 Jun;4(6):787-92.

# Dante Genomics

## Impact on the human health



We accelerate science to save more lives



Dante leverages expertise in science and healthcare to create **customized genomic analysis processes** that have an impact on people's lives.

## About Us

Dante Genomics is a leading global genomics and precision medicine company working to accelerate science to deliver better healthcare and ultimately, save lives with a more human approach to health.

Our mission is to leverage whole genome sequencing technology and variant interpretation, so that you can make informed decisions about your health and your future.

## Contact Info

Get in touch with us today!  
Contact us:



[customer.support@dantelabs.com](mailto:customer.support@dantelabs.com)

or visit our website framing the qr code or searching [dantegenomics.com](http://dantegenomics.com)

## Why Choose Dante Genomics?

### Actionable Reports



We provide actionable reports to support decision making.

### Dedicated Customer Success Team



A dedicated B2B customer support team at your complete disposal to support you throughout the analysis process.

### Scale and Expertise



Leverage our scale and expertise in whole genome sequencing and access attractive economics.

### Global

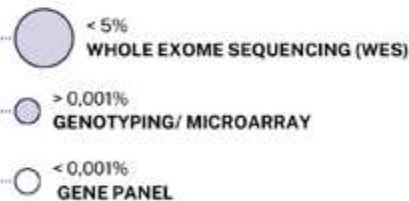
We cover countries across the globe, with an end-to-end service, including free return logistics, APIs and customer success teams

## Whole Genome Sequencing

100%  
WHOLE GENOME SEQUENCING (WGS)

- Clinical Grade 30X
- Whole Genome Sequencing
- Evidence-based information from ClinVar database
- CE-IVD software for genetic analysis

% OF ANALYSED GENOME



## Personalized medicine needs personalized data

The impact of genomics is changing the fundamental rules in the Healthcare and Pharma industries for both companies and individuals.

Dante Genomics is changing the rules providing:



End-to-End Genomic Solutions



Software and Genetic Reports



Drug Development

Dante Genomics accelerates science to make informed health decisions and save lives.





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Geneyx Analysis è una piattaforma innovativa per l'interpretazione e la refertazione dell'analisi dei dati Next Generation Sequencing (NGS) di interi genomi (WGS), interi esomi (WES), pannelli genici e Cancer screening. GDPR compliant, marchiato CE e ad oggi anche IVDR

Il software è distribuito in esclusiva per tutta Italia da Elettrobiochimica S.r.l.



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We're rewriting  
the **diagnostics**  
timeline.



OM~~AEY~~

CTCGCGTTGTCGCGACAC  
GT **YOUR RESEARCH** GAT  
ACTOUR **SEQUENCING** T  
CG **YOUR SUCCESS** GGTG  
TGGGGTTCGACACCGTTC



Humanizing Genomics  
**macrogen** EUROPE





HUMAN  
GENOME  
MEETING  
2024

8-10 April 2024,  
Rome - Italy

# SCIENTIFIC PROGRAMME DAY-BY-DAY





# MONDAY, 8 APRIL

09:00 -  
10:45

LUCCHESI ROOM  
MINERALOGIA

Educational workshop 1 | Knowledge and Skills for  
Multi-disciplinary Genomic Healthcare

*Dhavendra Kumar*

*Edward Tobias*

*Carolyn Applegate*

*Annie Hasan*

*Andreas Laner*

*Julie Makani*

*With case presentations from Sherifa Ahmed, Alison McEwen, Angela Solano, Zilfalil Bin Alwi, Charles Wray, and others*

PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO

Educational workshop 2 | Variant Interpretation / Variant Curation

*Andreas Laner*

ORGANI COLLEGIALI  
ROOM RETTORATO

Educational workshop 3 | All about Tools (OMIM, ClinGen, GA4GH tools, GenCC, AllOfUs, Matchmaker Exchange)

*Ada Hamosh*

*Erin Riggs*

*Marina Di Stefano*

*Peter Goodhand*

*Carolyn Applegate*

MULTIMEDIALE ROOM  
RETTORATO

Educational workshop 4 | Pangenomes Workshop

*Karen Miga*

*Simon Heumos*

*Xian Chang*

*Jean Monlong*

10:45 -  
11:30

Coffee break & Exhibition at Viale Gobetti

11:30 -  
12:45

LUCCHESI ROOM  
MINERALOGIA

Workshop 1 continued

PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO

Workshop 2 continued

ORGANI COLLEGIALI  
ROOM RETTORATO

Workshop 3 continued

MULTIMEDIALE ROOM  
RETTORATO

Workshop 4 continued

12:45 -  
14:00

Lunch & Exhibition at Viale Gobetti



14:00 -  
14:30

PLENARY 1  
AULA MAGNA  
RETTORATO

**Opening**

*Ada Hamosh, Johns Hopkins University, US*  
*Juergen Reichardt, James Cook University, Australia*  
*Giuseppe Novelli, Tor Vergata University of Rome, Italy*  
**Invited Authorities**

14:30 -  
15:05

*Mike Snyder, Stanford University, US*  
"Disrupting healthcare using deep data and remote monitoring"

PLENARY 1  
AULA MAGNA  
RETTORATO

**Symposium 1 - Precision Health**

Chair: *Bruno Dallapiccola, Bambino Gesù Hospital, Italy*

15:10 -  
15:35

*Francesca Forzano, NHS, UK*  
"Precision Screening?"

15:35 -  
16:00

*Marc Sultan, Roche, Switzerland*  
"Applications of genomics technologies in early drug development at Roche"

16:00 -  
16:25

**DANTE GENOMICS**  
*Nikil Sudarsan, King's College Hospital, UAE*  
"Case of Chronic granulomatous disease"  
"A Peek into Future of Genomics in practice"

PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO

**Symposium 2 - Reproductive Genetics**

Chair: *Fiorella Gurrieri, Campus Biomedico, Italy*

15:10 -  
15:35

*Diana Bianchi, NIH, US*  
"The NIH IDENTIFY study: a prospective evaluation of pregnant women with prenatal cfDNA sequencing results that suggest maternal malignancy"

15:35 -  
16:00

*Liborio Stuppia, University of Chieti-Pescara, Italy*  
"Epigenetics and human reproduction"

16:00 -  
16:25

**REVVITY**  
*Madhuri Hegde, Revvity*  
"Ensuring newborns recognize the benefits of genomic medicine"

16:25 -  
17:25

**Exhibition & Coffee Break at Viale Gobbetti**  
**Poster Session at Museo dell'Arte Classica | Lettere e Filosofia**

17:25 -  
17:40

PLENARY 1  
AULA MAGNA  
RETTORATO

**Introduction to HUGO & HUGO Committee Activities**

HUGO Past, Present, and Future  
*Walter Bodmer, UK*





17:40 -  
17:48

PLENARY 1  
AULA MAGNA  
RETTORATO

**HUGO Committees Introduction**

HUGO Committee on Ethics, Law and Society

*Donrich Thaldar, South Africa*

.....

17:48 -  
17:56

HUGO Gene/Disease Specific Database Advisory Council

*Ivo Fokkema, Netherlands*

.....

17:56 -  
18:04

HUGO Variant Nomenclature Committee

*Ivo Fokkema, Netherlands*

*Marina Di Stefano, Italy*

.....

18:04 -  
18:12

HUGO Pathogenicity

*Melissa Cline, US*

*Rosemary Ekong, UK*

.....

18:12 -  
18:20

HUGO Forum

*Juergen Reichardt, James Cook University, Australia*

.....

18:25 -  
20:30

Welcome Reception & Exhibition at Viale Gobbetti





# TUESDAY, 9 APRIL

PLENARY 1  
AULA MAGNA  
RETTORATO

## Symposium 3 | Gene Therapy

Chair: **Paolo Gasparini**, University of Trieste, Italy

09:00 -  
09:25

**Alessandro Aiuti**, San Raffaele Telethon Institute for Gene Therapy, Italy  
"Hematopoietic stem cell gene therapy for rare inborn errors of immunity and metabolism"

09:25 -  
09:50

**Luigi Naldini**, Vita-Salute San Raffaele University, Italy  
"Advanced engineering of hematopoiesis by gene editing strategies"

09:50 -  
10:15

### Oral Presentation

**Roni Hahn**, Tel Aviv University, Israel  
"Genetic therapy for hearing loss and vestibular dysfunction"

**Valentina D'Agostino**, Department of Pediatric Hematology and Oncology and of Cell and Gene Therapy, Bambino Gesù Children's Hospital IRCCS, Rome, Italy  
"Old diseases - new methods: beta-thalassemia erythroid cellular model using a base editing approach"

PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO

## Symposium 4 | Non-coding RNA genes

Chair: **Karen Avraham**, Tel Aviv University, Israel

09:00 -  
09:25

**Roderic Guigo**, CRG, Spain  
"GENCODE: Uncovering and cataloging the transcriptional complexity of the human and mouse genomes"

09:25 -  
09:50

**Tim Coorens**, Broad Institute, US  
"Variation and regulatory mechanisms of the small RNA transcriptome across human tissues"

09:50 -  
10:15

### Oral Presentation

**Beatrice Bodega**, INGM, Milan, Italy, Dept. of Biosciences, University of Milan, Milan, Italy  
"Transcripts containing Retrotransposable Elements play a regulatory role in modulating the functions of Tumor Infiltrating Lymphocytes"

**Chi Way Yip**, Center for Integrative Medical Sciences, RIKEN, Yokohama, Japan  
"Identification of novel transcript models by cap-trap full-length CDNA sequencing unveil transcription properties of non-coding RNA"

10:15 -  
11:00

Exhibition & Coffee Break at Viale Gobbetti  
Poster Session at Museo dell'Arte Classica | Lettere e Filosofia

11:00 -  
11:15

PLENARY 1  
AULA MAGNA  
RETTORATO

## Special Session

GA4GH Introduction  
**Peter Goodhand**, Canada

11:15 -  
11:35

PLENARY 1  
AULA MAGNA  
RETTORATO

## Special Session

HUGO Education Committee Introduction  
**Dhavendra Kumar**, UK



**PLENARY 1  
AULA MAGNA  
RETTORATO**

**Symposium 5 | Genetics of Complex Diseases**

Chair: **Massimo Gennarelli**, University of Brescia, Italy

11:35 -  
12:00

**Nicole Soranzo**, Human Technopole, Italy  
"High-resolution genomic analyses in human population"

12:00 -  
12:25

**Naomi Wray**, University of Queensland, Australia  
"Genetics of psychiatric vs gastrointestinal disorders"

12:25 -  
12:50

**Oral Presentation**

**Ammira Al-Shabeeb Akil**, Population Genetics and Genomic Medicine, Sidra Medicine, Doha, Qatar  
"Genetic factors in type 1 diabetes among the Qatari population: identification of HLA genotypes associated with enhanced or reduced risk of disease onset"

**Federica Santonastaso**, Genomics Research Centre, Human Technopole, Milan, Italy  
"Dissecting the biomedical and genetic structure of a southern italian cohort: the moli-sani study"

**PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO**

**Symposium 6 | Rare Diseases**

Chair: **Vincenzo Nigro**, Vanvitelli University, Italy

Co-Chair : **Anne Bowcock**, Mount Sinai, US

11:35 -  
12:00

**Steven Laurie**, CNAG, Spain  
"Solve-RD, Pan-European co-operation elevates the diagnostic rate in the most challenging rare disease cases"

12:00 -  
12:25

**William Gahl**, NIH, US  
"Rare and undiagnosed diseases: discoveries and Insights from the NIH undiagnosed diseases program"

12:25 -  
12:50

**Oral Presentation**

**Lara Guerrieri**, Department of Biomedicine and Prevention, Tor Vergata University, Rome, Italy  
"Characterization and aging assessment of human induced pluripotent and mesenchymal stem cells in MDPL syndrome"

**Annalaura Torella**, Precision Medicine, Università della Campania Luigi Vanvitelli, Napoli, Italy, Telethon Institute of Genetics and Medicine, Pozzuoli, Italy  
"A systematic approach for thousand severe unsolved pediatric conditions: results from the Telethon Undiagnosed Disease Program"

12:50 -  
14:00

**Lunch & Exhibition at Viale Gobbetti**

14:00 -  
14:30

**PLENARY 1  
AULA MAGNA  
RETTORATO**

**Corporate Symposium**

NEGEDIA

**Davide Cacchiarelli**, Telethon Institute of Genetics and Medicine, Italy  
"Bridging the gap between advanced sequencing technologies and clinical diagnostics: A history of the Telethon Research Applications"

14:30 -  
15:00

**Corporate Session**

THERMO FISHER

**Walter Ricciardi**, Cattolica University & Chair of the Horizon Europe Mission on Cancer, Italy  
"Present and future of Predictive Genomics in Italy and Europe"







14:00 -  
14:30

PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO

### Corporate Session

#### 4 BASES

**Prof. Eloisa Arbustini**, Center for Inherited Cardiovascular Diseases, IRCCS Fondazione Policlinico San Matteo, Pavia  
"How sequencing can impact precision medicine in a clinical context"

14:30 -  
15:00

### Meet the Expert Session

#### ILLUMINA

**Ivo Gut**, CNAG Barcelona  
**Pietro Gatti**, Illumina  
"A closer look at the role of genomics in advancing precision medicine: the '1+ Million Genomes' initiative"

14:00 -  
14:30

MULTIMEDIALE ROOM  
RETTORATO

### Corporate Session

#### TOMA ADVANCED BIOMEDICAL ASSAYS

**Riccardo Manca**, TomaLab  
"Health scenario on approaches to medical genetics"

**Annamaria Galiotta**, TomaLab  
"Technological approach and future of omics sciences in genetics"

**Elide Spinelli**, Fondazione Poliambulanza Hospital Brescia, Italy  
"Rare genetic disorders: medical geneticist's approach"

14:30 -  
15:00

### Corporate Session

#### OXFORD NANOPORE TECHNOLOGIES

**Alberto Magi**, Department of Information Engineering, University of Florence  
"Exploring cancer epigenomic evolution with PoreMeth2"

14:00 -  
14:30

ORGANI COLLEGIALI  
ROOM RETTORATO

### Meet the Expert Session

#### VERITAS

**Vincenzo Cirigliano**, Veritas  
**Luis Izquierdo**, Veritas  
**Michael Sandberg**, The London Genetics Centre, UK  
"Elective Genome Screening: its role in clinical practice and general health checks"

14:30 -  
15:00

### Meet the Expert Session

#### NOSTOS GENOMICS

**Valentina Ferradini**, Tor Vergata University of Rome, Italy  
"Reducing complexity in variant interpretation with AION: the experience of Tor Vergata Hospital"

15:00 -  
15:25

PLENARY 1  
AULA MAGNA  
RETTORATO

### Symposium 7 | Computational Biology & AI

Chair: **Juergen Reichardt**, James Cook University, Australia

**Alistair Forrest**, University of Western Australia, Australia  
"Spatial transcriptomics reveals discrete tumour microenvironments and autocrine loops within ovarian cancer subclones"

15:25 -  
15:50

**Susanna Zucca**, enGenome, Italy  
"Advancing genomic variant interpretation with artificial intelligence"





15:50 -  
16:15

### Oral Presentation

**Piero Luca Mazzacuva**, Department of Engineering, University Campus Bio-Medico di Roma, Roma, Italy, IBIOM-CNR, Bari, Italy  
"Profiling A to I RNA editing via TCN-based classifier"

**Giuseppe Giovanni Nardone**, 1 Department of Medicine, Surgery and Health Sciences, University of Trieste, Trieste, Italy  
"Optimizing Structural Variant Calling: towards a robust and reliable detection from Whole Genome Sequencing (WGS)"

PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO

### Symposium 8 | Genetics of Infectious Diseases

Chair: **Giuseppe Novelli**, Tor Vergata University of Rome, Italy

15:00 -  
15:25

**Emmanuelle Jouanguy**, Laboratory of Human Genetics of Infectious Diseases, Necker branch, Imagine institute, Paris Cité University, Laboratory of Human Genetics of Infectious Diseases, Rockefeller branch, Rockefeller University  
"Genetic and immunological predisposition to infectious diseases"

15:25 -  
15:50

**Christian Happi**, Redeemer's University, Nigeria  
"Genomic surveillance and characterization of microbial threats facilitates early detection and containment of disease outbreaks in West Africa"

15:50 -  
16:15

### Oral Presentation

**Michela Murdocca**, Department of Biomedicine and Prevention, University of Rome Tor Vergata, Rome, Italy  
"Peptide-designed strategies to counteract the evolution of SARS-COV-2 variants"

**Mark Seielstad**, Institute for Human Genetics, University of California San Francisco, San Francisco, USA  
"Genomic applications to global health: examples from tuberculosis and hepatitis B"

16:15 -  
17:15

Exhibition & Coffee Break at Viale Gobbetti  
Poster Session at Museo dell'Arte Classica | Lettere e Filosofia

PLENARY 1  
AULA MAGNA  
RETTORATO

### Symposium 9 | Cancer Genomics

Chair: **Maurizio Genuardi**, Catholic University, Italy

17:15 -  
17:40

**Serena Nik-Zainal**, University of Cambridge, UK  
"Whole cancer genomes: Insights and updates"

17:40 -  
18:05

**John Burn**, Newcastle University, UK  
"Lynch syndrome: A common rare disease, hiding in plain sight, the UK experience"

18:05 -  
18:30

### Oral Presentation

**Marcello Salvi**, Tigem, Napoli, Italy, Dieti, Unina, Italy  
"Multimodal genomic analysis unveils FGFR4 polymorphism and STAT3 pathway activation in cancer of unknown primary"

**Carla Debernardi**, Department of Medical Sciences, Unit of Genomic Variability and Complex diseases, University of Turin, Turin, Italy  
"Breast cancer risk polygenic score optimisation through a novel SNP selection algorithm"



PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO

**Symposium 10 | Single-cell Genomics / Spatial Genomics**

Chair: **Piero Carninci**, *Human Technopole, Italy*

**Kerstin Meyer**, *Wellcome Sanger Institute, UK*  
"From omics to hypothesis: Single cell and spatial profiling of the lung"

**Efrat Shema**, *Weizmann Institute of Science, Israel*  
"Decoding the epigenome for cancer research and diagnostics"

**Oral Presentation**

**Hagen Tilgner**, *Weill Cornell Medicine, New York City, USA*  
"Combined measurements of chromatin and splicing in thousands of individual cells reveals convergent and divergent patterns between both modalities in health and disease"

**Chung-Chau Hon**, *Center for Integrative Medical Sciences, RIKEN, Yokohama, Japan*  
"A single-cell atlas OH transcribed CIS-regulatory elements in human genome"

**Gala Dinner at Terrazza Caffarelli**

17:15 -  
17:40

17:40 -  
18:05

18:05 -  
18:30

20:00 -  
22:30





# WEDNESDAY, 10 APRIL

09:00 -  
09:30

PLENARY 1  
AULA MAGNA  
RETTORATO

## HUGO African Prize

**Leon Mutesa**, University of Rwanda, Rwanda  
"Intergenerational and epigenetic effects of trauma and PTSD"

09:30 -  
10:30

Exhibition & Coffee Break at Viale Gobbetti  
Poster Session at Museo dell'Arte Classica | Lettere e Filosofia

10:30 -  
11:10

PLENARY 1  
AULA MAGNA  
RETTORATO

## HUGO Chen Awards

**Chen Award for Distinguished Academic Achievement in Human Genetic and Genomic Research**  
**Peter Visscher**, University of Queensland, Australia "Height as a model trait in human complex trait genetics"

11:10 -  
11:40

**Chen Award of Excellence**  
**Zornitza Stark**, University of Melbourne, Australia  
"Accelerating rare disease diagnosis"

11:40 -  
13:00

Lunch & Exhibition at Viale Gobbetti

13:00 -  
13:40

PLENARY 1  
AULA MAGNA  
RETTORATO

## Corporate Symposium

QIAGEN

**Michela Bulfoni**, Azienda Sanitaria Universitaria Friuli Centrale, Italy  
"Benefits of integrating targeted next-generation sequencing (NGS) panels into molecular diagnostics of solid tumors"

13:40 -  
14:15

## Oral Presentation Session

Precision Health

Chair: **Giacomo Frati**, Sapienza University of Rome, Italy  
Co-Chair: **Federica Sangiuolo**, Tor Vergata University, Italy

### Oral presentation

**Bianca De Nicolo**, Department of Medical and Surgical Sciences, University of Bologna, Bologna, Italy  
"Donor-derived cell free DNA is associated with antibody-mediated rejection in patients with heart transplantation"

**Josefin Bjurling**, Department of Immunology, Genetics and Pathology, Science for Life Laboratory, Uppsala University, Uppsala, Sweden  
"Contribution of mosaic loss of chromosome Y to sex bias in idiopathic pulmonary fibrosis"

**Beatrice Spedicati**, Department of Medicine, Surgery and Health Sciences, University of Trieste, Trieste, Italy, Institute for Maternal and Child Health - I.R.C.C.S. "Burlo Garofolo", Trieste, Italy  
"When time matters: application of high-throughput sequencing technologies in critically-ill infants admitted to neonatal and pediatric intensive care units (NICU/PICU)"



13:00 -  
13:40

PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO

### Oral Presentation Session

#### Reproductive Genetics

Chair: **Antonio Novelli**, Bambino Gesù Hospital, Italy

Co-chair: **Paola Grammatico**, Sapienza University of Rome, Italy

#### Oral Presentation

**Jan Diblík**, GENNET, Prague, Czech Republic  
"Extended carrier screening in clinical practice"

**Alenka Hodžić**, Clinical Institute of Genomic Medicine, University Medical Centre Ljubljana, Ljubljana, Slovenia  
"Genetic testing for monogenic etiology of male infertility contributes to the clinical diagnosis of men with severe idiopathic male infertility"

#### Challenges in Assessing Pathogenicity of Variants

**Lorenzo Vaccaro**, Telethon Institute of Genetics and Medicine (TIGEM), Pozzuoli (NA), Italy, Department of Translational Medicine (DISMET), University of Naples "Federico II", Naples, Italy  
"A novel deep mutational scanning approach to dissect at single cell level the molecular bases of genetic diseases"

**Giulia Pianigiani**, Institute for Maternal and Child Health - I.R.C.C.S. "Burlo Garofolo", Trieste, Italy  
"Assessing the functional role of novel gene mutations associated to hearing loss with human iPSC-derived Inner Ear Organoids"

13:40 -  
14:15

### Oral Presentation Session

#### Pangenomes and Genomic Diversity

Chair: **Carla Jodice**, Tor Vergata University, Rome, Italy

Co-Chair: **Patrizia Malaspina**, Tor Vergata University, Rome, Italy

#### Oral presentation

**Giuliana Giannuzzi**, Department of Biosciences, University of Milan, Milan, Italy  
"Agap duplicons underlie structural diversity at chromosome 10Q11.22"

**Elisabetta Casalone**, Department of Medical Sciences, University of Turin, Turin, Italy  
"NIG: Network for Italian Genomes"

13:00 -  
13:40

MULTIMEDIALE ROOM  
RETTORATO

### Corporate Session

#### MGI & EUROCLONE

**Antonio Grimaldi**, Telethon Institute of Genetics and Medicine (TIGEM), Armenise/Harvard Laboratory of Integrative Genomics, Italy, University of Naples Federico II, Italy  
"Benchmarking of short reads sequencing in clinical genomics applications"

13:40 -  
14:15

### Corporate Session

#### ROCHE

**Claudia Cesario**, Bambino Gesù Hospital, Italy  
"Development of an automated workflow in the molecular characterization of Vascular anomalies"

14:15 -  
14:40

PLENARY 1  
AULA MAGNA  
RETTORATO

### Symposium 11 | Transgenerational Inheritance / Epigenetics

Chair: **Antonio Pizzuti**, Sapienza University of Rome, Italy

**Jill Fahrner**, Johns Hopkins University, US  
"Mendelian disorders of the epigenetic machinery and the use of DNA methylation profiling in diagnosis and discovery"

14:40 -  
15:05

**Simona Giunta**, Sapienza University of Rome, Italy  
"Complete diploid isogenomic reference human genomes for epigenetic precision"





15:05 -  
15:30

PLENARY 1  
AULA MAGNA  
RETTORATO

**Oral Presentation Session**

**Alessia Mauri**, Department of Biomedical and Clinical Sciences, University of Milan, Milan, Italy, Center of Functional Genomics and Rare Diseases, Buzzi Children's Hospital, Milan, Italy  
"Ketogenic diet therapies for drug-resistant epilepsy might affect ion channels activity through the combination of both epigenetic changes and splicing events"

**Daria Borodko**, Laboratory of Molecular Genetic Modelling of Inflammaging, Institute of General Pathology and Pathophysiology, Moscow, Russia  
"Differential methylation in ancient and modern people suggest early appearance of chronic inflammatory disease"

14:15 -  
14:40

PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO

**Symposium 12 | Challenges in Assessing Pathogenicity of Variants**

Chair: **Borut Peterlin**, University Medical Center Ljubljana, Slovenia

**Alan Rubin**, University of Melbourne, Australia  
"Enabling clinical translation of high-throughput mutagenesis Data with MaveDB"

14:40 -  
15:05

**Doug Speed**, Aarhus University, Denmark  
"Computational tools to improve our understanding of the biological basis of complex traits"

15:05 -  
15:30

**Melissa Cline**, UCSC, US  
"Engaging researchers with patients"

15:30 -  
16:20

**Exhibition & Coffee Break at Viale Gobbetti**  
**Poster Session at Museo dell'Arte Classica | Lettere e Filosofia**

16:20 -  
16:45

PLENARY 1  
AULA MAGNA  
RETTORATO

**Symposium 13 | Pangenomes and Genomic Diversity**

Chair: **Ada Hamosh**, Johns Hopkins University, US

**Karen Miga**, UCSC, US  
"The human pangenome project: Creating a reference that better represents human global genetic diversity"

16:45 -  
17:10

**Charles Lee**, The Jackson Laboratory, US  
"What have we learned from sequencing the entire human Y chromosome?"

17:10 -  
17:35

**Alfredo Coppa**, Sapienza University of Rome, Italy  
"All roads lead to Rome: DNA from bones reveals migration and diversity in ancient Rome"

16:20 -  
16:45

PLENARY 2  
GINESTRA ROOM  
CHIMICA  
CANNIZZARO

**Symposium 14 | Microbiome and Metagenomics**

Chair: **Giorgia Grotto**, University of Trieste, Italy

**Mireia Valles-Colomer**, Universitat Pompeu Fabra, Barcelona, Spain  
"The microbiome and health: Focusing on interpersonal transmission"

16:45 -  
17:10

**Serena Sanna**, Institute for Genetic and Biomedical Research, Italy  
"Human genome-microbiome interactions in health and diseases"

17:10 -  
17:35

**Oral Presentation Session**

**Gloria Sala**, Department of Health Sciences, University of Eastern Piedmont, Novara, Italy  
"Role of inherited predisposition and intestinal microbiota in colorectal carcinogenesis"

**Francesco Mugnai**, 4bases Italia Srl, Pavia, Italy  
"A clinical ready, long reads solution for sequencing human microbiota"





17:35 -  
18:00

PLENARY 1  
AULA MAGNA  
RETTORATO

Poster Prize

18:00 -  
18:20

PLENARY 1  
AULA MAGNA  
RETTORATO

Introduction to HGM2025 in Durban, South Africa

*Veron Ramsuran, University of KwaZulu-Natal, South Africa*

18:20 -  
18:35

PLENARY 1  
AULA MAGNA  
RETTORATO

Closing



# COVID

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## Editor-in-Chief

**Prof. Dr. Giuseppe Novelli**  
University of Rome Tor Vergata,  
Rome, Italy

## Message from the Editor-in-Chief

You are invited to contribute a research article or a comprehensive review for consideration and publication in *COVID* (ISSN 2673-8112). *COVID* is published in open access format—research articles, reviews and other content are released on the internet immediately after acceptance. The scientific community and the general public have unlimited free access to the content as soon as it is published. With the efforts of our professional team and the great potential of *COVID* topics, the impact factor of this journal will dramatically increase in the coming years. We would be pleased to welcome you as one of our authors.

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Clinical Treatment

Treatment Development

Public Health

Healthcare and COVID Complications

Global Impact

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**Booth  
#20**

**MGI**  
*EuroGene*

**Join our seminar**

## **Benchmarking of short reads sequencing in clinical genomics**

When: 10 April

Time: 13.00 - 13.30

Venue: Parallel Hall

Speaker: Dr. Antonio Grimaldi, Telethon Institute of Genetics and Medicine (TIGEM), Italy

**Want to try out the  
tech?**

Visit us at booth #10

**Join our symposium**

## **See what you're missing – nanopore sequencing for cancer research**

Date: Tuesday, 9 April 2024

Time: 2:30 – 3:00 pm CEST

Location: Hall Parallel 1, Rectorate Building

Speaker: Alberto Magi, Associate Professor, University of Florence



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➔ **Meet the team at booth 6-7** ➔



Let's talk with  
Negedia Chief Scientific Officer,  
Prof. Davide Cacchiarelli:  
**"Bridging the gap between  
advanced sequencing technologies  
and clinical diagnostics:  
a history of the Telethon research  
applications"**

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**April 9**



**14:00**

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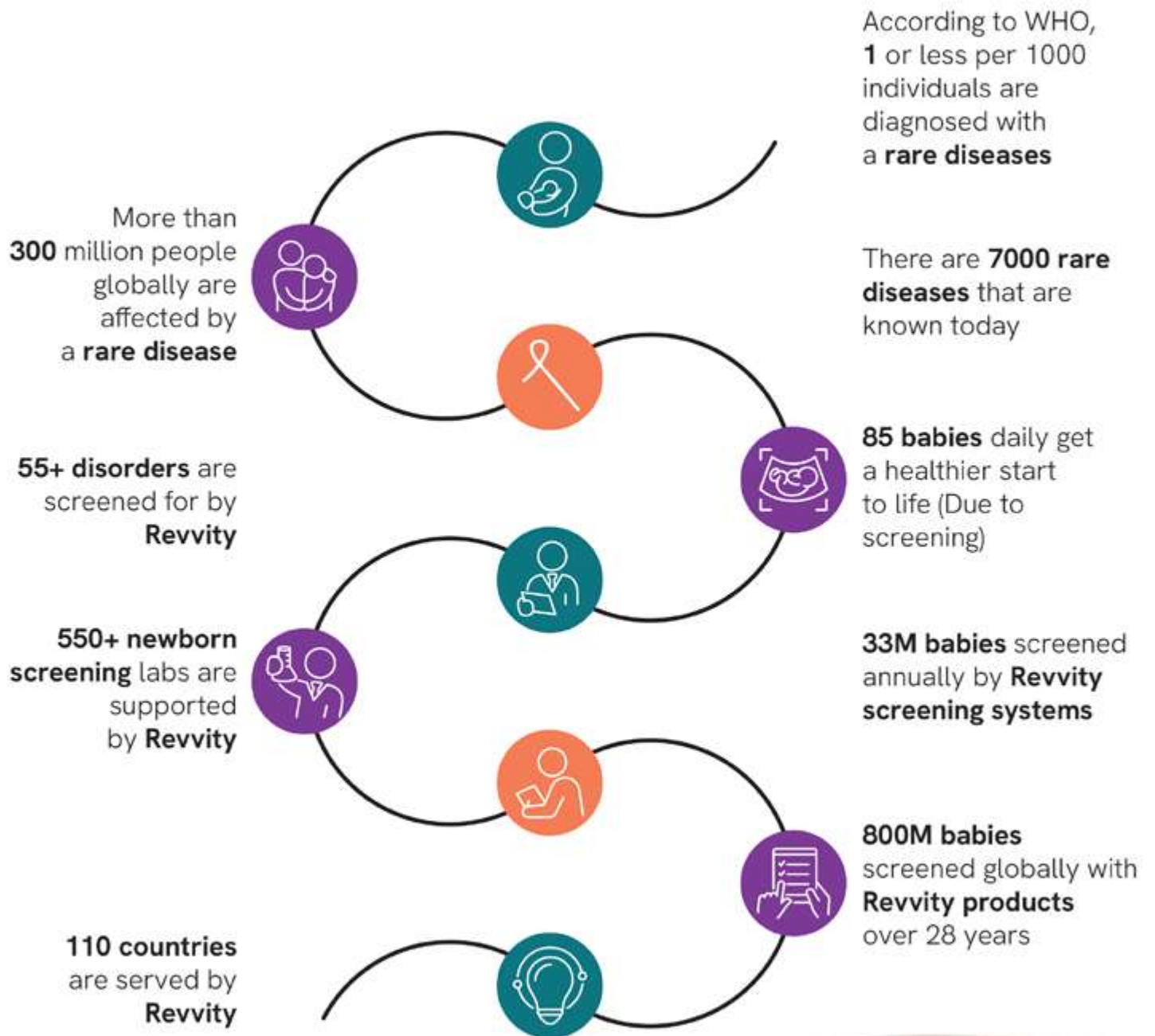
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Aiuti Alessandro – Italy  
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Zucca Susanna – Italy



# General information

## VENUE

Sapienza University of Rome  
Piazzale Aldo Moro, 5 – 00185 Rome

## HGM WEBSITE

[www.hugo-hgm2024.org](http://www.hugo-hgm2024.org)

## ORGANIZING SECRETARIAT

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REGISTRATION FEES	Early Bird by January 31	Standard by February 29	Late / on site from March 1
HUGO Member and non Member	€450	€550	€700
Students/Early Career (<5 yrs post doc)/Residents and Young Scientists (under 28 years)/Retired	€200	€300	€450
Developing Countries	€100	€100	€100
Industry	€590	€690	€790
SIGU,ESHG, FNOB member	€405	€495	€630

*All prices are VAT included*

Participants may be asked to present their ID to prove their student and post-doc status or a signed self-report to prove their membership to ESHG, SIGU or FNOB.

HGM2024 uses the World Bank's Classification of Countries to categorize the fees.

Individuals living and working in low-income and lower-middle income countries must provide proof of nationality for that country and proof

of employment/residence in that country.

## REGISTRATION FEE INCLUDES

Participation in all scientific sessions  
Congress bag

Printed materials of the Congress

An Invitation to the Welcome Reception

Lunches and coffee breaks for 3 days

Entrance to the Exhibition area



## **SOCIAL EVENT**

HGM social dinner at Terrazza Caffarelli will be held on April 9, 2024 at 8.00 p.m and can be purchased on HGM website.

Terrazza Caffarelli

Piazza Caffarelli, 4

<https://maps.app.goo.gl/JznRY1w9tQQfL7fK9>

For the entrance, it's requested to show the badge and the copy of the dinner receipt.

## **BADGE**

Each regularly registered participant and exhibitor will be provided with a badge that must be exhibited at all times for the admission to the HGM meeting rooms and HGM exhibition area.

## **CERTIFICATE OF ATTENDANCE**

Certificates of attendance will be issued at the end of the event to all registered participants who request them to the Organizing Secretariat by email at [secretariat@hugo-hgm2024.org](mailto:secretariat@hugo-hgm2024.org)

## **CATERING SERVICES**

Catering services will be provided in the exhibition area according to the HGM programme. It's requested to show the badge.

## **POSTER SESSION**

Poster sessions will be organized in the poster area.

The presenting authors are requested to stay up-to-date with the HGM programme and poster schedule and to stay in front of their poster during the session.

Poster size maximum: 70 cm base x 100 cm high.

Each poster will have to be affixed by the presenting author before the beginning of the session and to be removed at the end. The Organizing Secretariat will not be responsible for uncollected posters.

## **RESPONSIBILITY AND INSURANCE**

The Organizers of HGM do not assume any responsibility for personal injury or loss / damage to goods belonging to participants or exhibitors during or after the event.

## **USE OF MOBILE PHONE, FILMING AND POSTS ON SOCIAL MEDIA**

The use of mobile phones in the meeting rooms is prohibited. It is also prohibited the filming of the speeches and posting on social media without express permission.



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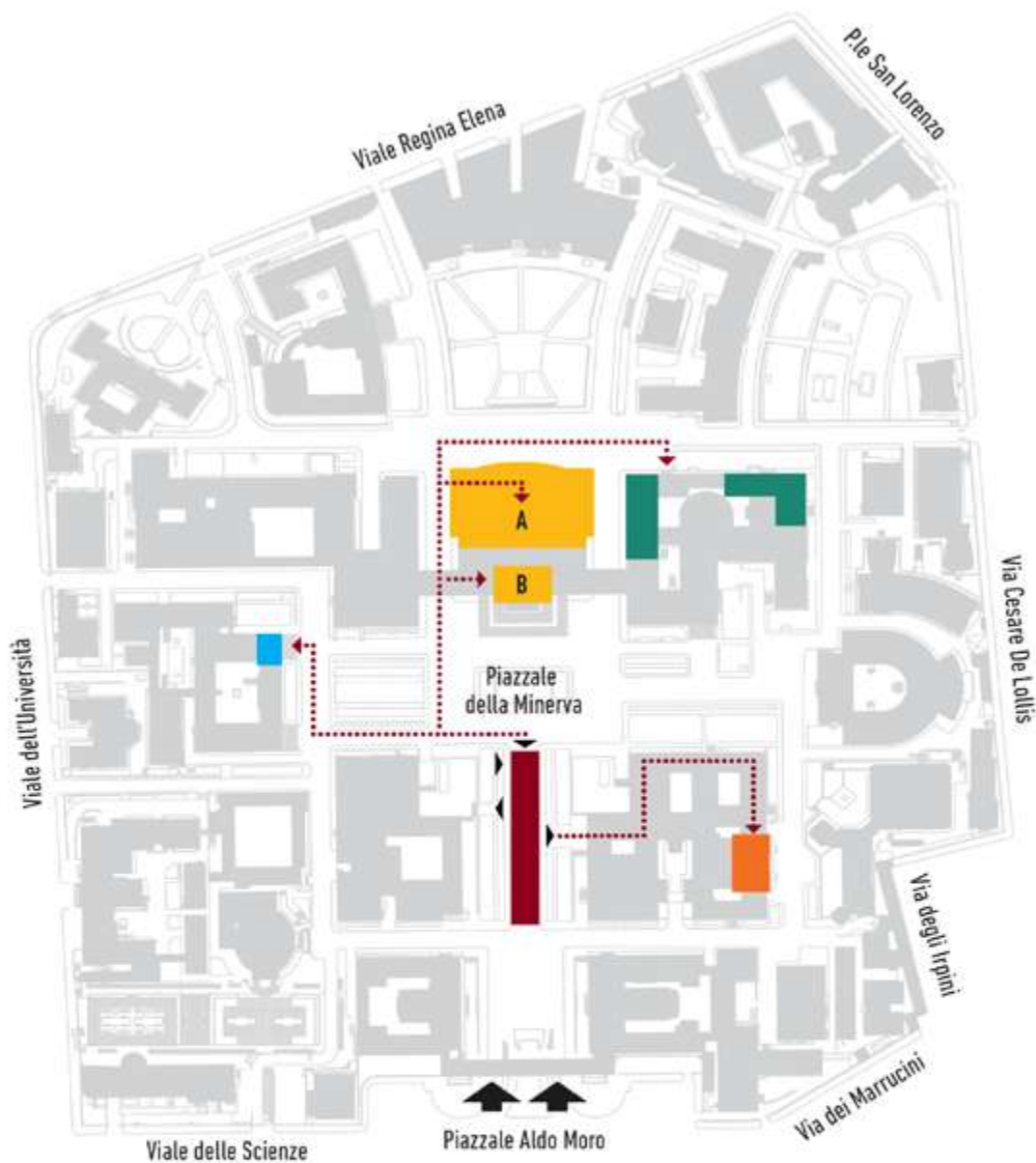
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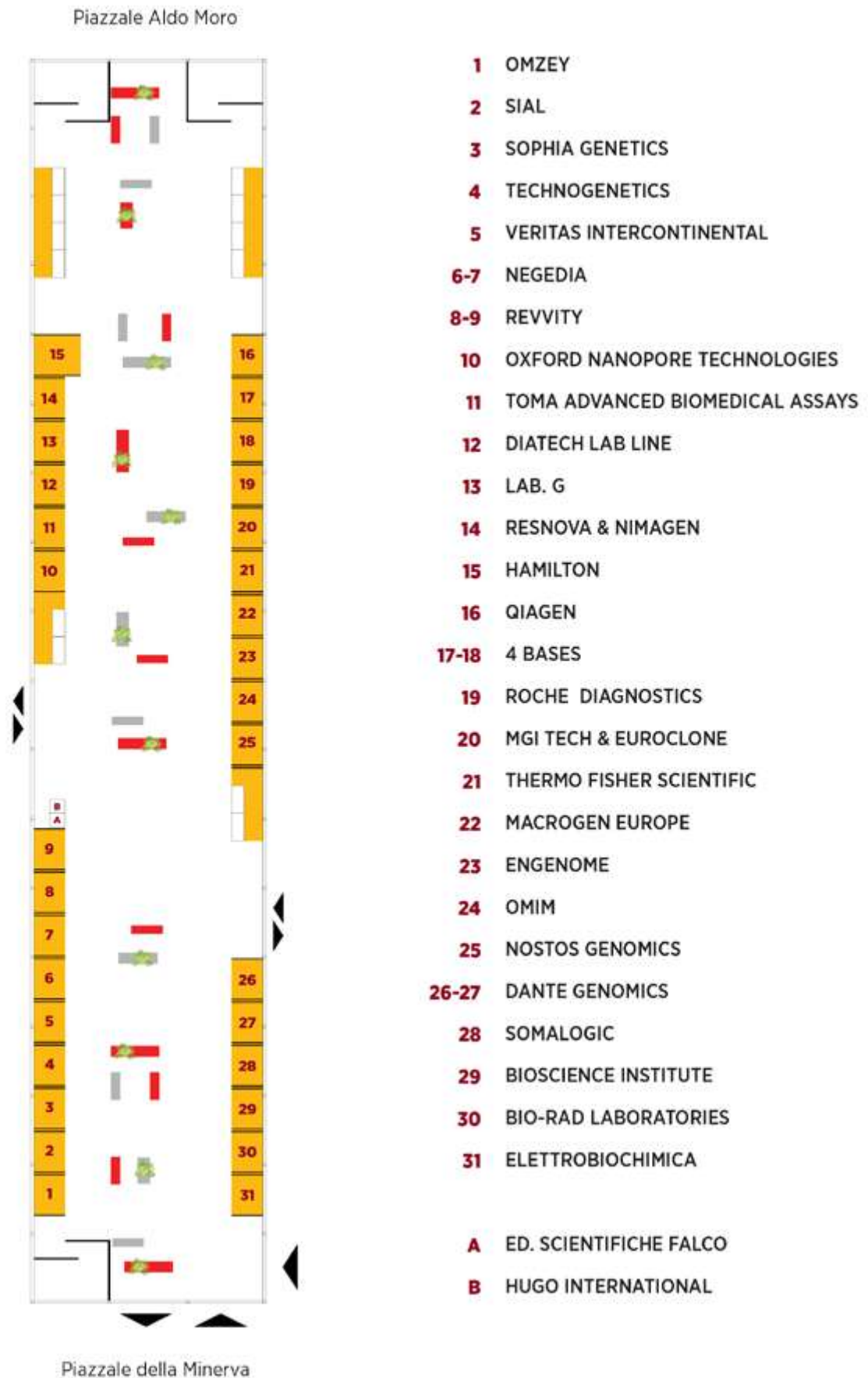
# Venue plan



- Rettorato A (CU001)**  
 Foyer: registration area  
 Aula Magna: plenary room 1
- Rettorato B (CU001)**  
 Aula Organi Collegiali: workshop / parallel room  
 Aula Multimediale: workshop / parallel room
- Chimica "Cannizzaro" (CU014)**  
 Aula Ginestra: plenary room 2  
 and workshop room
- Tensostucture Viale Piero Gobetti**  
 Exhibition / catering area
- Mineralogia (CU005)**  
 Aula Lucchesi: workshop room
- Museo dell'Arte Classica / Lettere e Filosofia (CU003)**  
 Poster area



# Exhibition area plan



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## PLATINUM



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