HUMAN GENOME ORGANISATION
HUGO

HUMAN GENOME MEETING 2024

8-10 April 2024, Rome - Italy

SCIENTIFIC PROGRAMME

Under the patronage of

Consiglio Nazionale delle Ricerche · ESHG · FNOB · Ordine dei Biologi · Sapienza Università di Roma · Tor Vergata

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Patronages

Under the patronage of

Consiglio Nazionale delle Ricerche

ESHG

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Adjunct Professor, Department of Pharmacology, University of Nevada, Reno, NV, USA

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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

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4bases

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.**

Introducing the *new* CFX Opus Real-Time PCR Systems

Equipped with the reliable features you know and expect from Bio-Rad, including differentiated Bio-Rad optics and CFX Maestro Software, plus improvements in design, functionality, and connectivity.

Integrated with the new BR.io cloud platform for experimental setup and data management. It's the perfect combination of everything you expect and everything you want for the modern lab.

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BIO-RAD

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	2022 Usage
4.5 - 2-year Impact Factor	510,313 downloads
4.9 - 5-year Impact Factor	1,940 Altmetric mentions

Editorial Board

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**
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**

* 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**

BMC
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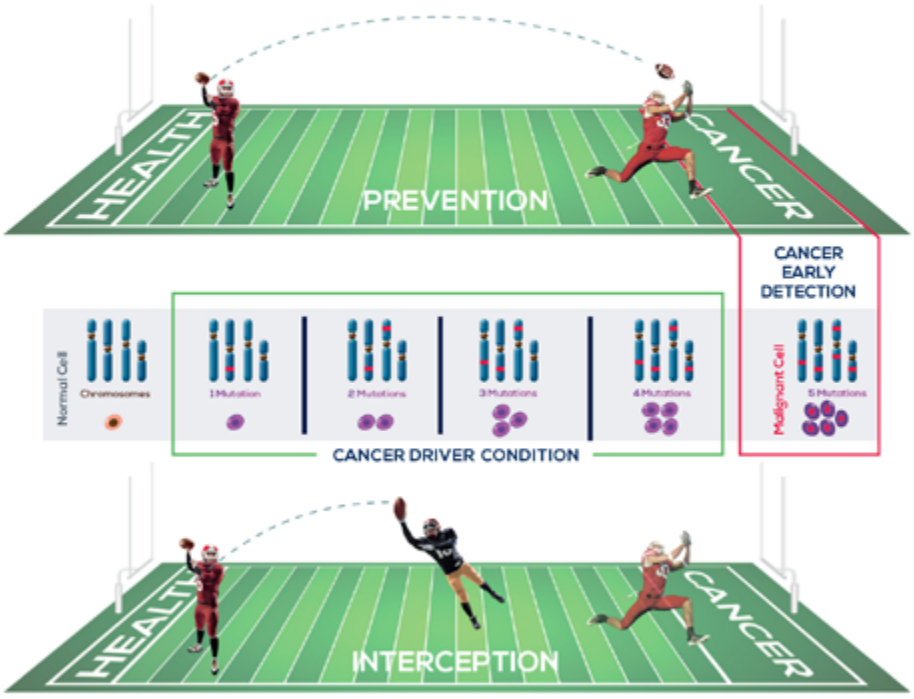
CANCER DRIVER INTERCEPTION

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.

BIOSCIENCE INSTITUTE

dante genomics

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

or visit our website framing the qr code or searching **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

- < 5% **WHOLE EXOME SEQUENCING (WES)**
- > 0,001% **GENOTYPING/ MICROARRAY**
- < 0,001% **GENE PANEL**

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.

JuliaOmix SUITE

We're rewriting the diagnostics timeline.

OMEey

YOUR RESEARCH
OUR SEQUENCING
YOUR SUCCESS

Humanizing Genomics
macrogen EUROPE

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 Lanear

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

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 Autorities

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*

PLENARY 1
AULA MAGNA
RETTORATO

17:40 - 17:48 — **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 transcrpit 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

PLENARY 1
AULA MAGNA
RETTORATO

14:00 - 14:30

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 Galietta, *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"

PLENARY 1
AULA MAGNA
RETTORATO

Symposium 7 | Computational Biology & AI

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

15:00 - 15:25

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 tubercolosis 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*

17:15 - 17:40

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

17:40 - 18:05

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

18:05 - 18:30

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"

20:00 - 22:30

Gala Dinner at Terrazza Caffarelli

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 patiens 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-throughout 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 contirbutes to the clinical diagnosis of men with severe idiophatic 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"

PLENARY 1
AULA MAGNA
RETTORATO

Symposium 11 | Transgenerational Inheritance / Epigenetics

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

14:15 - 14:40

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, Institutre of General Pathology and Pathophysiology, Moscow, Russia*
"Differential methylation in ancient and modern people suggest early appearance of chronic inflammatory disease"

PLENARY 2
GINESTRA ROOM
CHIMICA
CANNIZZARO

Symposium 12 | Challenges in Assessing Pathogenicity of Variants

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

14:15 - 14:40

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

PLENARY 1
AULA MAGNA
RETTORATO

Symposium 13 | Pangenomes and Genomic Diversity

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

16:20 - 16:45

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"

PLENARY 2
GINESTRA ROOM
CHIMICA
CANNIZZARO

Symposium 14 | Microbiome and Metagenomics

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

16:20 - 16:45

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

an Open Access Journal by MDPI

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.

Scope

Human or Animal Coronaviruses

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Treatment Development

Public Health

Healthcare and COVID Complications

Global Impact

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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

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

Save your seat

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NEGEDIA

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Innovating the processes of research and diagnosis of genetic diseases by providing the best performing solutions based on **Next Generation Sequencing**.

This is the mission of Negedia, a company born out of the **Telethon Foundation**'s desire to transfer the experience gained during its 30-year commitment to the fight against rare genetic diseases into a medical genetics laboratory of the highest scientific and technological level, open to the entire national and international clinical and medical community.

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"

SEE YOU AT NEGEDIA PRESENTATION!

Main hall, Rectorate Building

April 9 **14:00**

Negedia S.r.l. | Via Campi Flegrei 34 - 80078 Pozzuoli (NA)
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Empowering lives: Revvity's commitment to rare disease awareness

According to WHO, **1** or less per 1000 individuals are diagnosed with a **rare diseases**

More than **300** million people globally are affected by a **rare disease**

There are **7000 rare diseases** that are known today

55+ disorders are screened for by **Revvity**

85 babies daily get a healthier start to life (Due to screening)

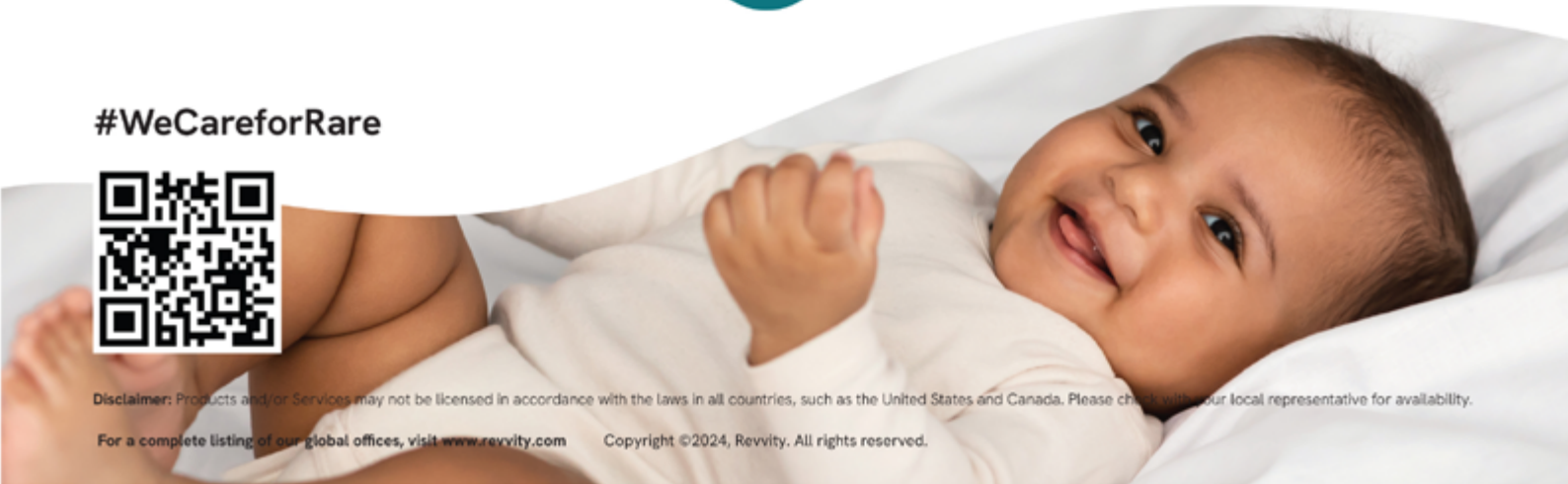
550+ newborn screening labs are supported by **Revvity**

33M babies screened annually by **Revvity screening systems**

800M babies screened globally with **Revvity products** over 28 years

110 countries are served by **Revvity**

#WeCareforRare



Speaker & Chairman

Ahmed Sherifa – Egypt
Applegate Carolyn – US
Aiuti Alessandro – Italy
Arbustini Eloisa - Italy
Avraham Karen – Israel
Bianchi Diana – US
Bin Alwi Zilfalil – Malaysia
Bodmer Walter – United Kingdom
Bowcock Anne – US
Bulfoni Michela – Italy
Burn John – United Kingdom
Cacchiarelli Davide – Italy
Carninci Piero – Italy
Cesario Claudia – Italy
Chang Xian – US
Cirigliano Vincenzo – Veritas
Cline Melissa – US
Coorens Tim – US
Coppa Alfredo – Italy
Dallapiccola Bruno – Italy
Di Stefano Marina – Italy
Ekong Rosemary – United Kingdom
Fahrner Jill – US
Ferradini Valentina - Italy
Fokkema Ivo – Netherlands
Forrest Alistair – Australia
Forzano Francesca – United Kingdom
Frati Giacomo – Italy
Gahl William – US
Galietta Annamaria – Italy
Gasparini Paolo – Italy
Gatti Pietro – Illumina
Gennarelli Massimo – Italy
Genuardi Maurizio – Italy
Girotto Giorgia – Italy
Giunta Simona – Italy
Goodhand Peter – Canada
Grammatico Paola – Italy
Grimaldi Antonio – Italy
Guigo Roderic – Spain
Gurrieri Fiorella – Italy
Gut Ivo – Spain
Hamosh Ada – US
Happi Christian – Nigeria
Hasan Annie – India
Hegde Madhuri – Revvity
Heumos Simon – Germany
Izquierdo Luis – Spain
Jodice Carla – Italy
Jouanguy Emmanuelle – France
Kumar Dhavendra – United Kingdom
Laner Andreas – Germany
Laurie Steven – Spain
Lee Charles – US
Magi Alberto – Italy
Makani Julie – United Kingdom
Manca Riccardo – Italy
Malaspina Patrizia – Italy
McEwen Alison – Australia
Meyer Kerstin – United Kingdom
Miga Karen – US
Monlong Jean – France
Mutesa Leon – Rwanda
Naldini Luigi – Italy
Nigro Vincenzo – Italy
Nik-Zainal Serena – United Kingdom
Novelli Antonio – Italy
Novelli Giuseppe – Italy
Peterlin Birut – Slovenia
Pizzuti Antonio – Italy
Ramsuran Veron – South Africa
Reichardt Juergen – Australia
Ricciardi Walter - Italy
Riggs Erin – US
Rubin Alan – Australia
Sandberg Michael – United Kingdom
Sangiuolo Federica – Italy
Sanna Serena – Italy
Snyder Mike – US
Shema Efrat – Israel
Soranzo Nicole – Italy
Speed Doug – Denmark
Spinelli Elide – Italy
Stark Zornitza – Australia
Stuppia Liborio – Italy
Sudarsan Nikil – Dante Genomics
Sultan Marc – Switzerland
Thaldar Donrich – South Africa
Tobias Edward – United Kingdom
Valles-Colomer Mireia – Italy
Visscher Peter – Australia
Wray Charles – United States of America
Wray Naomi – Australia
Zucca Susanna – Italy

General information

VENUE

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

HGM WEBSITE

www. hugo-hgm2024.org

ORGANIZING SECRETARIAT

done!

DONE SRL
Via Monte delle Gioie, 1
00199 Rome Italy
info@hugo-hgm2024.org
www.hugo-hgm2024.org

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 Carreer (<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 Cuntries 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

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.

HUMAN GENOME MEETING 2024

SOPHiA GENETICS™

ABOUT

SOPHiA GENETICS is a cloud-based software company that leverages the power of artificial intelligence to aid healthcare professionals in the fight against cancer and rare diseases. SOPHiA GENETICS is on a mission to make precision medicine the gold standard of care globally by providing users with access to a rich collection of diverse data from patients on a global scale.

SOPHiA GENETICS provides a platform, SOPHiA DDM™, that brings together patient data to speed the diagnostics process; assist physicians in making informed, data-driven treatment plans; and support cancer and rare disease researchers. In addition, SOPHiA GENETICS' technology supports the biopharma industry by helping accelerate clinical trials.

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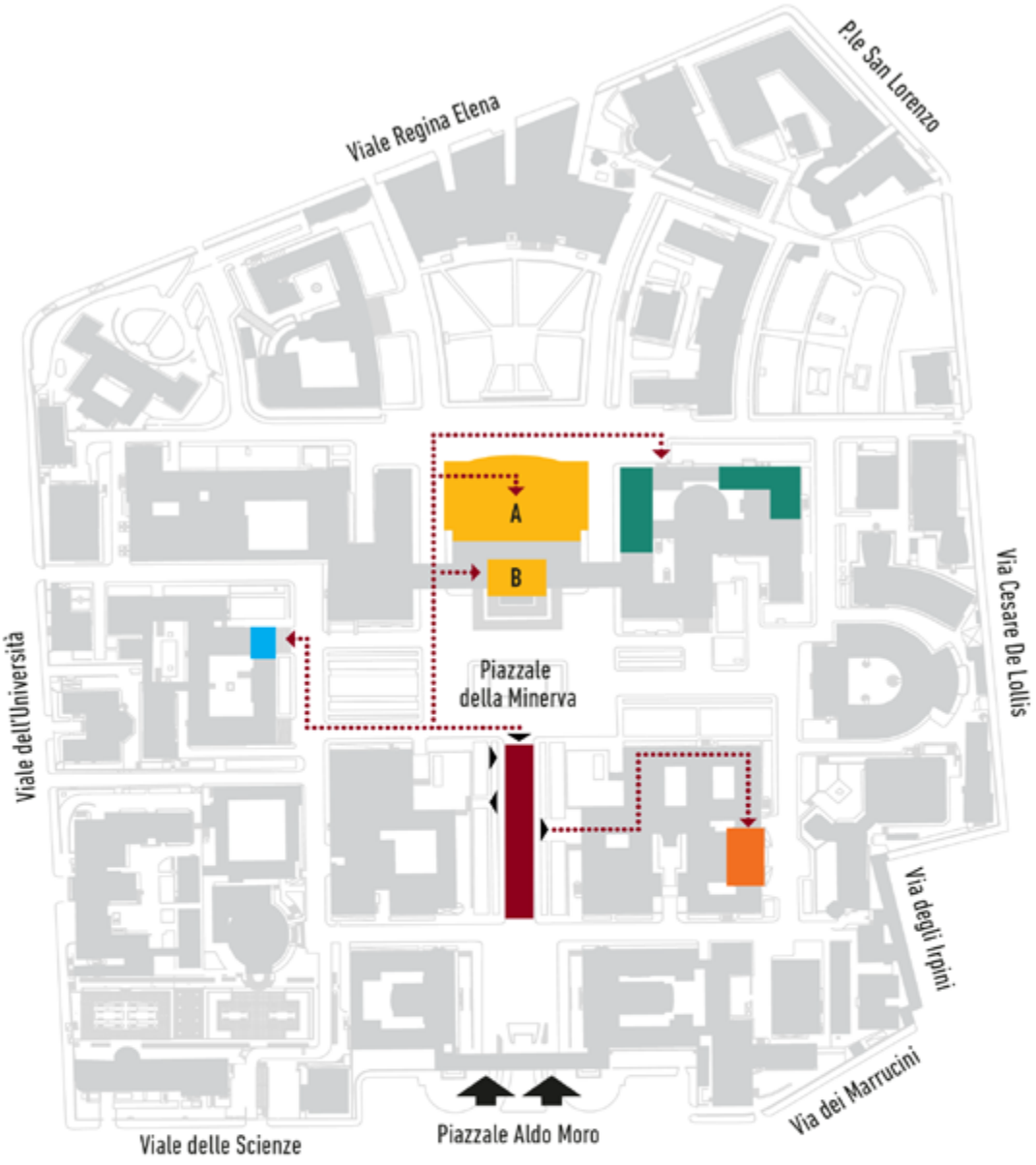
Venue plan



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

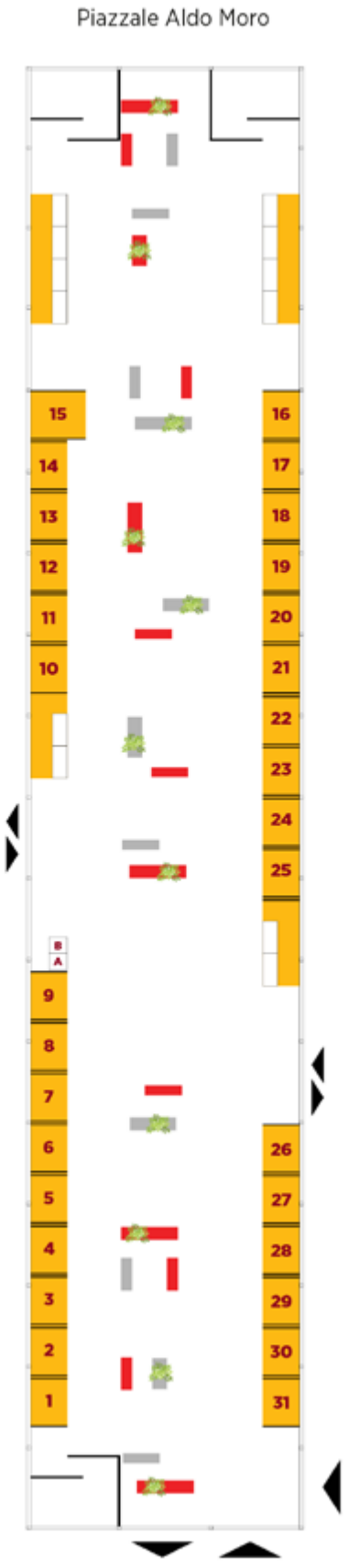
Chimica "Cannizzaro" (CU014)
Aula Ginestra: plenary room 2
and workshop room

Tensostructure 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



1	OMZEY
2	SIAL
3	SOPHIA GENETICS
4	TECHNOGENETICS
5	VERITAS INTERCONTINENTAL
6-7	NEGEDIA
8-9	REVVITY
10	OXFORD NANOPORE TECHNOLOGIES
11	TOMA ADVANCED BIOMEDICAL ASSAYS
12	DIATECH LAB LINE
13	LAB. G
14	RESNOVA & NIMAGEN
15	HAMILTON
16	QIAGEN
17-18	4 BASES
19	ROCHE DIAGNOSTICS
20	MGI TECH & EUROCLONE
21	THERMO FISHER SCIENTIFIC
22	MACROGEN EUROPE
23	ENGENOME
24	OMIM
25	NOSTOS GENOMICS
26-27	DANTE GENOMICS
28	SOMALOGIC
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30	BIO-RAD LABORATORIES
31	ELETTROBIOCHIMICA
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