

# EUROPEAN HUMAN GENETICS CONFERENCE 2025

# Hybrid Conference Allianz MiCo | Milan – Italy | May 24 – 27



# EUROPEAN SOCIETY OF HUMAN GENETICS PROGRAMME https://2025.eshg.org/ #e

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### GENERAL COMMITTEES - BOARD - ORGANISATION

### **European Society of Human Genetics**

**The ESHG** was established on March 15, 1967 by a small group of outstanding European Geneticists (European Journal of Human Genetics: 3, 63-64, 1995). The decision which led to the founding of the ESHG was taken at an informal meeting of European participants at the III<sup>rd</sup> International Congress of Human Genetics in Chicago in September 1966. Membership was open; the first annual membership fee was \$6. The founding chairman was J. Mohr who was secretary-general of the society until 1991.

**The Society** has an Executive Board (comprising the officers of the Society) who report to the Governing Board, which consists of about 30 members. According to the statutes, each year a sufficient number of Board members are elected to guarantee a balanced group in terms of gender, speciality and country representation. Up to half of the vacant positions can be proposed by the Board. The General Assembly, taking place at the conference, needs to confirm the proposals.

The Board has set up several committees to oversee aspects of the Society organisation, and appoints the chairmen. A full activity report is presented to members at the General Assembly at the annual European Human Genetics Conference.

There is an Annual Meetings Committee which is responsible for all the practical and legal arrangements for the European Human Genetics Conference. The scientific programme is arranged by the Scientific Programme Committee.

### **ESHG Office**

European Society of Human Genetics

c/o Vienna Medical Academy Alser Strasse 4, 1090 Vienna, AT https://2025.eshg.org/ T: +43 1 405 13 83 39 F: +43 1 407 82 74 E: office@eshg.org membership@eshg.org

### **European Human Genetics Conference 2025**

Conference Organisation and Abstract Management

ESHG 2025 Conference Office Wiener Medizinische Akademie GmbH Jana Wacht, Lydia Schnedl Alser Strasse 4, 1090 Vienna, AT T: +43 1 405 13 83 35 / 14 F: +43 1 407 82 74 E: conference@eshg.org https://2025.eshg.org/

### Exhibition, Sponsoring and Corporate Sessions & Presentations

Rose INTERNATIONAL Exhibition Management and Congress Consultancy bv Jantie de Roos, Flora van Laer P.O. Box 93260 2509 AG The Hague, NL T: +31 70 383 8901 F: +31 70 381 8936 E: eshg@rose-international.com

### **Hotel Accommodation**

MiCodmc P.le Carlo Magno 1 20149 Milan – Italy T: +39 02 4997 6275 E: eshg2025.hotels@micodmc.it





### **Future European Human Genetics Conferences**

European Human Genetics Conference 2026 Gothenburg, Sweden June 13 – 16, 2026

European Human Genetics Conference 2027 Rotterdam, Netherlands June 12 – 15, 2027

European Human Genetics Conference 2028 Helsinki, Finland June 17 – 20, 2028

### CME Credits

As in previous years, the European Genetics Conference has applied for accreditation from the European Accreditation Council of Continuing Medical Education (EACCME®). The accreditation is still pending at the time of printing this programme. Please see ESHG conference website for the latest updates: https://2025.eshg.org/myconference/cme-credits/

Through an agreement between the Union Européenne des Médecins Spécialistes and the American Medical Association, physicians may convert EACCME<sup>®</sup> credits to an equivalent number of AMA PRA Category 1 Credits<sup>™</sup>. Information on the process to convert EACCME<sup>®</sup> credit to AMA credit can be found at https://edhub.ama-assn.org/pages/applications. Live educational activities, occurring outside of Canada, recognised by the UEMS-EACCME<sup>®</sup> for ECMEC<sup>®</sup>s are deemed to be Accredited Group Learning Activities (Section 1) as defined by the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada.



## GENERAL HYBRID CONFERENCE PLATFORMS

### Virtual Conference Platform

All registered participants have access to the ESHG 2025 virtual conference platform until Nov. 30, 2025. On the platform participants will find:

- Full conference programme incl. available corporate sessions & presentations
- The option to create their own personalised programme
- Live stream of all conference sessions
   Live stream of corporate sessions as indicated on the Virtual Platform
- Hybrid posters and E-Posters in digital form
- Access on-demand content
- · Profiles of speakers and Early Career Award Candidates

Participants can access the platform using the same e-mail address and password as used for registration. Scan the QR code to access the virtual conference platform.

### The ESHG 2025 Conference App

Do you always want to be up-to-date? The ESHG Society App will guide you through the programme day by day or by session type, will make available profiles of speakers and poster presenters and help you to find exhibitors by name or by services provided. Add papers or entire sessions to your personal mobile calendar, receive push messages with important reminders.

Available for iOS and Android in your App and Play Stores. Search for European Society of Human Genetics.

Download the mobile app https://2025.eshg.org/myconference/conference-app/

### Sequencing Square in the Exhibition

Formerly known as the 'Livestream Area', this has now been re-named to 'Sequencing Square'. Besides the Best Poster presentations, this area will function as a meeting and networking hub as well as hosting some programme during exhibition opening hours such as Workshops, Get2Gethers and Corporate Presentations.

### **Overflow Area**

The Cube Lounge on level 2 (link to Auditorium) will be used as an overflow area if needed. Participants will be informed through an app notification when it is in use.

### **IMPORTANT NOTICE**

Please note that taking pictures or filming during the sessions is allowed, unless explicitly requested otherwise by the presenter.







#### **PROGRAMME - SATURDAY MAY 24** 6

ROOM	GOLD PLENARY	AUDITORIUM	SPACE 1+2	BROWN 3
LEVEL	2	3	0	2
08.30 - 10.00	S01 Long-read genomes in health and disease S01.1 L. Milani S01.2 T. Pastinen S01.3 W. Timp	E01 Single cell genomics and diseases E01.1 A. Aschenbrenner E01.2 S. Bizzotto	WO1 The art of writing, reading, understanding and interpreting genetic reports J. Baptista, D. Vears, J. O'Byrne, SA. Lynch	SO2 Viral Mimicry in Cancer S02.1 C. Plass S02.2 D. de Carvalho S02.3 B. Greenbaum
10.00 - 10.30		Coffee Break (E	xhibition Hall 4)	I
10.30 - 12.00	C01 Improving the	CO2 GWAS: bigger,	CO3 Developmental	CO4 Cardiovascular
	diagnostic yield of rare diseases C01.1 S. Walker C01.2 C. Simoes Amaro* C01.3 B. van der Sanden* C01.4 M. Ek* C01.5 Y. Chen* C01.6 S. Facchini	faster, stronger           C02.1 N. Kumasaka           C02.2 Z. Yang*           C02.3 A. Luckett*           C02.4 H. Zheng*           C02.5 N. Baya*           C02.6 E. Abner	disorders C03.1 Y. Kovas C03.2 I. Ambrosetti* C03.3 M. Morleo C03.4 S. Yatsenko C03.5 J. Poulter C03.6 A. Reymond	disorders C04.1 B. Reversade C04.2 M. Patel C04.3 I. Gandin C04.4 N. Telis C04.5 H. Manikpurage* C04.6 P. Møller
12.00 - 13.45	Poster V	iewing - Lunch Break - E (Exhibition Hall 4)	khibition	12.15 - 13.15 ESHG Podcast
13.45 - 14.15	PLO Welcoming Address			
14.15 - 14.30	PL1 ELPAG Award PL1.1 S. Metcalfe			
14.30 - 16.00	PL2 Opening Plenary Session PL2.1 K. Miga PL2.2 S. McCarroll PL3.3 J. Krause			
16.00 - 16.30	Fruit B	Break, Free Poster Viewin	g, Exhibition (Exhibition	Hall 4)
16.30 - 18.00	PL3 What's New? PL3.1 E. Morini PL3.2 V. Yumiceba* PL3.3 T. Naert PL3.4 T. de Bitter* PL3.5 F. Taylan PL3.6 D. Kasperaviciute			
18.00 - 18.30	Coffee	Break, Free Poster Viewi	ng, Exhibition (Exhibition	n Hall 4)
18.30 - 20.00	CO9 Genomewide approaches for rare diseases (09.1 D. Pava C09.2 A. McGuigan* (09.3 G. Devadoss Gandhi* (09.4 F. Lecoquierre* (09.5 J. Y. Ha* (09.6 J. Ellingford	C10 New technologies and tools C10.1 J. Mudge C10.2 J. K. Kim C10.3 P. Keukeleire* C10.4 A. Daponte* C10.5 C. Kajba* C10.6 L. Jolly	C11 New genes and mechanisms in Intellec- tual Disability (11.1 K. Platzer (11.2 Y. Shi* (11.3 C. Pailler-Pradeau* (11.4 J. Bou-Rouphael* (11.5 J. Paccaud	C12 Decoding complex traits through molecular intermediates (12.1 A. Raveane (12.2 A. Argentieri* (13.3 A. Lopez de Lapuente Portilla (12.4 A. Ray (12.5 S. He (12.6 F. Küçükali*
		HG 2025   Milan – Italy   https:/		



# PROGRAMME - SATURDAY MAY 24 Z

For a complete overview of the Saturday Corporate Sessions go to page 20

0 SO3 Gaucher's and Parkinson's disease: partners in crime S03.1 R. Asselta	SPACE 4 0 WO3 GenEthics: confidentiality and privacy in genomic	SEQUENCING SQUARE HALL 4 LEVEL 2 / (IN-PERSON ONLY)	CORPORATE SESSIONS	ROOM LEVEL	
SO3 Gaucher's and Parkinson's disease: partners in crime SO3.1 R. Asselta	WO3 GenEthics: confidentiality and privacy in genomic		SESSIONS		
and Parkinson's disease: partners in crime S03.1 R. Asselta	confidentiality and privacy in genomic			00 20 10 00	
S03.2 S. Bandres Ciga S03.3 E. Sidransky	research and datasets T. Clancy, H. Carley, R. Horn, A. McNeill			08.30 - 10.00	
Coffee Break (Exhibition Hall 4)					
C06         Deep dive           into neurogenetics         06.1 S. Gu           (06.2 S. Saha*         06.3 N. Bertola *           (06.3 N. Bertola *         06.4 L. Bicknell           (06.5 M. Nesson-Dauphin*         06.6 G. Nicolas	C07 Metabolism and Mitochondria (07.1 M. Tarailo-Graovac (07.2 J. Soenksen* (07.3 E. Davis (07.4 A. Newman* (07.5 D. Ciotlos* (07.6 J. Christodoulou	CO8 Best Poster 1	10.00 - 11.15 Corporate Sessions	10.30 - 12.00	
GO1 12.15 - 13.15 Uniting European Young Initiatives in Human Genetics	Exhib	pition	12.15 - 13.30 Corporate Sessions	12.00 - 13.45	
				13.45 - 14.15	
				14.15 - 14.30	
				14.30 - 16.00	
uit Break, Free Post	er Viewing, Exhibiti	on (Exhibition Hall 4	4)	16.00 - 16.30	
				16.30 - 18.00	
Coffee Break, Free Poster Viewing, Exhibition (Exhibition Hall 4)					
statistical genetics approaches C14.1 M. Rivas C14.2 F. Wang* C14.3 J. Zeng C14.4 J. Hof* C14.5 S. Wang*	ative disorders of nerve and muscle (15.1 S. Mero (15.2 N. Nabavizadeh* (15.3 J. Parmar* (15.4 L. Dofash* (15.5 M. Nashabat*			18.30 - 20.00	
	Coffee CO6 Deep dive nto neurogenetics 106.2 S. Saha* 106.3 N. Bertola * 106.4 L. Bicknell 106.5 M. Nesson-Dauphin* 106.6 G. Nicolas GO1 12.15 - 13.15 Juiting European Indiana Genetics GO1 12.15 - 13.15 Juiting European Go1 12.15 - 13.15 Ju	R. Horn, A. McNeill         Coffee Break (Exhibition I         Cooffee Break, Free Poster Viewing         Poster Viewing         Exhibition         tit Break, Free Poster Viewing, Exhibition         Cooffee Break, Free Poster Viewing, Exhibition         tit Break, Free Poster Viewing, Exhibition         Cooffee Break, Free Po	R. Horn, A. McNeill         Coffee Break (Exhibition Hall 4)         Coffee Break (Exhibition Gravac         Coffee Break, Instead         Offee Break, Free Poster Viewing - Lunch Break - Exhibition (Exhibition Hall 4)         Interpean (Exhibition Hall 4)         Interpean (Exhibition Hall 4)         It Break, Free Poster Viewing, Exhibition (Exhibition Hall 4)         It Break, Free Poster Viewing, Exhibition (Exhibition Hall 4)         It also begenerative disorders of nerve and muscle         It also begenerative disor	303.3 E. Sidransky       R. Horn, A. McNeill         Coffee Break (Exhibition Hall 4)         C06       Deep dive nto neurogenetics 006.1 S. Gu       C07       Metabolism and Mitochondria 007.1 M. Tarailo-Gravvac 007.21. Soenksen*       C08       10.00 - 11.15         C06.1 S. Gu       C07.1 M. Tarailo-Gravvac 006.3 N. Bertola *       C07.3 L. Davis       Deep dive C07.4 N. Newman*       C07.5 D. Ciotlos*         C06.1 L. Bicknell       C07.5 D. Ciotlos*       C07.6 J. Christodoulou       Dester Viewing - Lunch Break - Exhibition (Exhibition Hall 4)       12.15 - 13.30         G01       12.15 - 13.15       Poster Viewing - Lunch Break - Exhibition (Exhibition Hall 4)       12.15 - 13.30         divid Buropean foung initatives       Poster Viewing, Exhibition Hall 4)       12.15 - 13.30         divid Break, Free Poster Viewing, Exhibition (Exhibition Hall 4)       Image: Component of the set	



# **PROGRAMME - SUNDAY MAY 25**

ROOM	GOLD PLENARY	AUDITORIUM	SPACE 1+2	BROWN 3	
LEVEL	2	3	0	2	
08.30 - 10.00	S04 Genetics at single cell resolution: spatial, temporal and morphology S04.1 J. Powell S04.2 O. Stegle S04.3 H. Kilpinen	W04 Long-read sequencing for beginners C. Gilissen, G. Santen, M. Delledonne, M. Eberle, A. Doran, W. Höps	W05 Structural chromosomal variants N. de Leeuw, A. Engwerda, R. Wijngaard, A. da Cruz	WO6 Nonsense- mediated mRNA decay: variant interpretation challenges J. Gecz, F. Millan, F. Lejeune, S. Lunke, L. Jolly	
10.00 - 10.30	Coffee	Break, Free Poster Viewi	ng, Exhibition (Exhibition	n Hall 4)	
10.30 - 12.00	C16 U R New - RNU-	C17 RNAseg for rare	C18 Large-scale and	C19 Genetic epi-	
	related disease discoveries C16.1 M. Quinodoz C16.2 S. Cuinat* C16.3 A. Torella C16.4 A. Santini* C16.5 A. Jackson C16.6 D. Greene	diseases C17.1 N. Pipko C17.2 S. Kumble* C17.3 N. Tan* C17.4 N. Jalkanen* C17.5 L. Vaccaro* C17.6 C. Africano*	multi-omic approaches to neuropsychiatric conditions (18.10. Quenez (18.2 C. Liao* (18.3 E. Assary* (18.4 A. Saparov (18.5 A. R. Marques* (18.6 T. Jensen	demiology: Powering prediction and precision (19.1 U. Marigorta (19.2 J. German* (19.3 L. Urpa* (19.4 C. Albiñana* (19.5 A. Man* (19.6 R. Mustafa*	
12.00 - 13.00		1	1	1	
	LUNCH, Free Poster Viewing, Exhibition (Exhibition Hall 4)				
13.00 - 14.00	Poster Viewing with Authors and coffee (Group A) (Exhibition/Poster Hall 4)				
14.00 - 14.15		Time to cho	ange rooms		
14.15 - 15.45	SOG Somatic muta- tions in normal tissues SOG.1 L. Harvey SOG.2 L. Forsberg SOG.3 K. Burns	WO9 Mutations we almost missed C. Gilissen, C. Ding, J. Baptista, G. Ravenscroft, V. Yépez, B. Van Der Sanden	S07 Genetics of Al-derived phenotypes 507.1 C. Glastonbury 507.2 N. Cai 507.3 J. Pirruccello	SO8 Genomics of big neuromuscular genes S08.1 S. Treves S08.2 M. Savarese S08.3 G. Ravenscroft	
15.45 - 16.00	Fruit B	Break, Free Poster Viewin	g, Exhibition (Exhibition	Hall 4)	
16.00 - 17.00	Poster Viewing with Authors and coffee (Group B) (Exhibition/Poster Hall 4)				
17.00 - 17.15		Time to cho	ange rooms		
17.15 - 18.45	S10 The non-coding genome: gene regulation and disease S10.1 M. Vollger S10.2 E. de Baere S10.3 M. Mhlanga	S11 Penetrance and Expressivity S11.1 R. Barbosa Matos S11.2 C. Auwerx S11.3 K. McGurk	EO3 Cancer Evolution E03.1 M. Tanić E03.2 M. Efremova	E04 Scalable proteomics empower human genetics and epidemiology E04.1 A. Butterworth E04.2 J. Schwenk	
2.02.00 🖶 🖬 🚺					



# PROGRAMME - SUNDAY MAY 25

### For a complete overview of the Sunday Corporate Sessions go to page 20

BROWN 1+2	SPACE 3	SPACE 4	SEQUENCING SQUARE HALL 4	CORPORATE	ROOM
2	0	0	LEVEL 2 / (IN-PERSON ONLY)	SESSIONS	LEVEL
E02 New treat- ment avenues E02.1 H. Zhou E02.2 C. Venditti	S05 Decoding actionability, severity, and utility in genomics S05.1 E. M. Bunnik S05.2 Y. Bombard S05.3 F. Boardman	W07 Implement- ing pharmacoge- netic testing into de clinics: let's debate C. Rodriguez-Antona, J. Yang, V. Pratt, J. McDermott, D. Müller, E. Cecchin, H. L. Wee		08.30 - 10.00 Corporate Sessions	08.30 - 10.00
Coffee Break, Free Poster Viewing, Exhibition (Exhibition Hall 4)					10.00 - 10.30
C20 Cancer Com- plexity: Variants, Therapies, and Al-Driven Insights C20.1 M. C. Roa Bravo C20.2 A. Nunes* C20.3 L. Bulić* C20.4 A. Brandão C20.5 J. Li* C20.6 l. Franco	C21 Prenatal and reproductive medicine C21.1 Y. Gao C21.2 P. Cao C21.3 S. Hoffman* C21.4 A. Bashamboo C21.5 F. R. Grati C21.6 W-J. Wang	C22 Immunology and Hematology C22.1 E. Cohen Barak C22.2 A. Lamarca C22.3 G. Sato C22.4 F. Diab C22.5 T. H. Heng* C22.6 E. Papalexi	C23 Best Poster 2		10.30 - 12.00
W08 12.15 - 13.45 Publishing genom- ics research	GO2 12.15 - 13.15 European Reference Networks: The time		12.15 - 12.45 Corporate Presentation	12.00 - 13.00 Corporate Sessions	12.00 - 13.00
A. McNeill, A. Niselle, J. Kist, S. Riedijk	and c	g with Authors offee tion/Poster Hall 4)	13.00 - 13.30 Corporate Presentation		13.00 - 14.00
		ime to change room	15		14.00 - 14.15
S09 Fetal thera- pies S09.1 L. Guilbaud S09.2 C. Götherström S09.3 H. Schneider	W10 Tools for clinical genome interpretation M. Holtgrewe, A. Lermine, S. Ossowski, P. Robinson, J. Fasham, M. Landrum	W11 Prioritizing the counselling in genetic counselling T. Wessels, A. Mendes, R. Moldovan, J. Austin, A. McEwen	GO3 14.15 - 15.45 The 1+MG/GDI/ Genome of Europe and All of Us Initiatives	14.15 - 15.45 Corporate Sessions	14.15 - 15.45
Fi	ruit Break, Free Post	ter Viewing, Exhibiti	ion (Exhibition Hall	4)	15.45 - 16.00
	wing with Authors o B) (Exhibition/Poste		G04 16.00 - 17.00 GenQA supporting the implementation of ISCN 2024		16.00 - 17.00
Time to change rooms					17.00 - 17.15
S12 Expanding data sources: tech- nical, ethical and legal considerations S12.1 G. Ertaylan S12.2 M. Kogut-Czarkowska S12.3 E. Rial-Sebbag	E05 Leukodys- trophies E05.1 A. Shukla E05.2 E. Giorgio	S13 Does genomic screening leave open the door to eugenics? S13.1 E. Parens S13.2 G. Cavaliere S13.3 A. Newson			17.15 - 18.45

#### **PROGRAMME - MONDAY MAY 26** 10

ROOM	GOLD PLENARY	AUDITORIUM	SPACE 1+2	BROWN 3		
LEVEL	2	3	0	2		
08.30 - 10.00	S14 Reassessing com- plex trait inheritance through eQTLs S14.1 H. Mostafavi S14.2 K. Alasoo S14.3 A. Claringbould	E06 Effects of Struc- tural Variants on the 3D Genome E06.1 M. Spielmann E06.2 T. Rausch	S15 Implementing gene editing S15.1 A. Cereseto S15.2 F. Locatelli S15.3 A. Auricchio	W12 Genomic Newborn Screening: learning from current programmes D. Kasperaviciute, A. Pichini, A. Ferlini, P. Tsipouras, W. Chung, Z. Stark, D. Vears, G. Gumus		
10.00 - 10.30	Coffee	n Hall 4)				
10.30 - 12.00	C24 Multiomics for	C25 Functional and	C26 Computational	C27 New Treatments		
	rare diseases (24.1 G. Bullich (24.2.J. Carrasco Zanini Sanchez* (24.3 D. Hock* (24.4 R. Shaath* (24.5 C. LaFlamme* (24.6 C. Thauvin	structural genomics (25.1 C. Gross (25.2 P. Haffener (25.3 S. Fornezza* (25.4 P. Clavell-Revelles* (25.5 A-L. Katzke* (25.6 V. Verhoeven	models in human genetics (26.1 E. Yeger-Lotem (26.2 C. Yakaboski* (26.3 R. Hofmeister* (26.4 Y. Li* (26.5 A. M. Vergani* (26.6 S. Londhe*	C27.1 X. Jeunemaitre C27.2 L. Fast* C27.3 A. Ziegler C27.4 E. Tasciotti C27.5 J. Baruteau C27.6 P. Prontera		
12.00 - 13.00		I	1			
	LUNCH, Free Poster Viewing, Exhibition (Exhibition Hall 4) M 12.15 - 13.15 ESHG General Assem					
13.00 - 14.00		oster Viewing with Autho and coffee				
	(Group C) (Exhibition/Poster Hall 4)					
14.00 - 14.15		Time to cho	ange rooms			
14.15 - 15.45	S17 Integrating big data and functional genomics S17.1 M. Claussnitzer S17.2 J. Yang S17.3 A. Morris	W15 Episignatures: utility and challenges C. Charbonnier Le Clézio, R. Weksberg, S. Choufani, M. Defrance	W15     Episignatures:     S18     Telomere-driven       utility and challenges     aging and disease       C. Charbonnier Le Clézio,     S18.1 J. Karlseder       R. Weksberg, S. Choufani,     S18.2 R. DePinho			
15.45 - 16.00	Fruit B	Break, Free Poster Viewin	g, Exhibition (Exhibition	Hall 4)		
16.00 - 17.00			h Authors and coffee tion/Poster Hall 4)			
17.00 - 17.15		Time to cho	ange rooms			
17.15 - 18.45	S20 Implementation of automated reanalysis of genomic data S20.1 Z. Stark S20.2 M. Plantinga S20.3 J. Buchanan	S21 Insights from con- sanguineous populations - Genomes of South Asia S21.1 H. Martin S21.2 H. Arsalan S21.3 P. Moorjani	E07 Bridging Genetics and Epigenetics in Human Diseases E07.1 B. Horsthemke E07.2 Y. Elgersma	EOB Mendelian Rand- omization - concepts and misconceptions E08.1 K. Fischer E08.2 S. Burgess		
			(2025 osba ora)			
三にやその	ESHG 2025   Milan – Italy   https://2025.eshg.org/					



PROGRAMME - MONDAY MAY 26

### For a complete overview of the Monday Corporate Sessions go to page 21

BROWN 1+2	SPACE 3	SPACE 4	SEQUENCING SQUARE HALL 4	CORPORATE	ROOM
2	0	0	LEVEL 2 / (IN-PERSON ONLY)	SESSIONS	LEVEL
W13 Dysmor- phology 2 S. Douzgou Houge, A. Bouman, P. Krawitz Q. Sabbagh*, M. Abdel-Hamid, F. Nazer*, A. Spano, E. Akbas*, H. Havriushenko, S. Carbonera*, R. Traberg	S16 Growth retardation: endo- crinologists meet geneticists S16.1 D. van der Kaay S16.2 I. Netchine S16.3 A. Dauber	W14 Responsible access to genomic data: who should keep the oversight? M. Shabani, Y. Moreau, R. Cook-Deegan, L. Marelli, D. Mascalzoni, J. LoTempio		08.30 - 10.00 Corporate Sessions	08.30 - 10.00
Ca	offee Break, Free Pos	ster Viewing, Exhibi	tion (Exhibition Hall	4)	10.00 - 10.30
C28 Congenital anomalies (28.1 A. Yeung (28.2 R. Oegema (28.3 C. De Leonibus (28.4 E. Pisan* (28.5 J. D. Gutiérrez-Ávila* (28.6 F. Cifarelli	C29 Late Break- ing Abstracts (29.1 Z. Fattahi (29.2 R. Harkness (29.3 S. Küry (29.4 K. Chundru (29.5 N. Whiffin (29.6 S. Giunta	C30 Emotional and social com- plexities in genetic counselling (30.1 M. Golas (30.2 T. Hoffmann* (30.3 T. Wainstein* (30.4 H. Dolling* (30.5 S. Pinto da Silva* (30.6 E. DeBortoli	C31 Best Poster 3		10.30 - 12.00
G05 12.15 - 13.15 Medical Genetics Professionals: what we can anticipate for the		Poster Viewing, hibition Hall 4)	12.15 - 12.45 Corporate Presentation	12.00 - 13.00 Corporate Sessions	12.00 - 13.00
Poster Viewing with Authors and coffee (Group C) (Exhibition/Poster Hall 4)					
	Т	ïme to change roon	15		14.00 - 14.15
S19 Lipids and diseases S19.1 P. De Camilli S19.2 C. Gehin S19.3 S. Clark	W17 UCSC and ENSEMBL genome browsers M. Haeussler, A. Mushtaq, J. Mudge, J. Allen, O. Austine-Orimoloye, J. Birgmeier	W18 Setting up your gene therapy clinical trial: dos and don'ts, tricks and tips L. Servais, N. Brunetti-Pierri, S. Milosevic, M. E. Bernardo, C. Martin	W19 Has social media broken scientific knowl- edge sharing? J. Fasham, A. Mushtaq, V. Rajopal, B. Talibani, R. Matos	14.15 - 15.45 Corporate Sessions	14.15 - 15.45
F	ruit Break, Free Pos	ter Viewing, Exhibit	ion (Exhibition Hall	4)	15.45 - 16.00
Poster Viewing with Authors and coffee (Group D) (Exhibition/Poster Hall 4)					16.00 - 17.00
Time to change rooms					17.00 - 17.15
S22 Rewriting cancer treatment and resistance S22.1 J. J. Yang S22.2 A. Vähärautio S22.3 A. Cox	S23 Rhopathies: Role of Rho GTPases and related genes beyond cancer S23.1 W. M. Bement S23.2 T. Millard	S24 Biomolecu- lar Condensates S24.1 M. Zhang S24.2 J. Ditlev S24.3 K. Strømgaard			17.15 - 18.45
SEED THE CON	S23.3 S. Schmidt				

ESHG 2025 | Milan – Italy | https://2025.eshg.org/

# 12 PROGRAMME - TUESDAY MAY 27

ROOM	GOLD PLENARY	AUDITORIUM	SPACE 1+2	BROWN 3
LEVEL	2	3	0	2
09.00 - 10.30	E09 Solving rare disease: 1 for all, all for 1 E09.1 K. M. Boycott E09.2 V. Yepez	S25 Fetal phenotyping S25.1 S. Thakur S25.2 I. Simcock S25.3 F. Dhombres	S26         Ultrapersonalised           medicine            S26.1 T. Yu            S26.2 C. Bupp            S26.3 S. Senn	S27 Chromosome X S27.1 E. Heard S27.2 B. Payer S27.3 M. Borensztein
10.30 - 11.00	Coffee I	Break (Balcony level 1, Cu	ube Lounge & Bar Miland	level 2)
11.00 - 12.30	C32 Beyond common variants: population-scale sequencing & CNV (32.1 S. Rubinacci (32.2 H. Wright* (32.3 G. Hawkes (32.4 G. Kalantzis* (32.5 A. Hanaya Alsuwaidi* (32.6 H. I. Kim	C33 Population genetics tracing evolution signatures (33.1 P. Dasmeh (33.2 MD. Zanoaga (33.3 K. Anastasiadou* (33.4 X. Han (33.4 K. Hal (33.6 I. Gallego Romero	C34 Advancing cancer prevention, detection and treatment (34.1 S. Tuveri (34.2 J. Burn (34.3 J. Lu* (34.4 V. B. Serio* (34.5 S. Pereira (34.6 G. Kungulovski	C35 Skeletal disorders C35.1 C. Chai Gadot C35.2 E. Bolmer C35.3 H. Lyubenova* C35.4 G. Imren* C35.5 E. Calpena C35.6 M. Doyard
12.30 - 13.30		Lunch (So	outh Hall)	
13.30 - 14.15	<b>PL4</b> Mendel Lecture PL4.1 K. Karikó			
14.15 - 15.00	PL5 ESHG Award Lecture PL5.1 D. MacArthur			
15.00 - 16.00	PL6 Awards & Closing			



# PROGRAMME - TUESDAY MAY 27

Session Types:

Plenary Session	Symposium	Concurrent Session
Educational Session	Get2Gether Session	Corporate Sessions & Presentations
Workshop		

### **IMPORTANT NOTICE:**

Please note that taking pictures or filming during the sessions is allowed, unless requested otherwise by the presenter. Persons who will not observe this rule will be excluded from the session by the chairpersons.



Session Type Description



### **ESHG Early Career Award Candidates**

The Scientific Programme Committee has shortlisted presenters for the **ESHG Early Career Awards (ECA).** The committee will judge the finalists' presentations during the conference. The following awards will be presented to the winners in the closing ceremony on Tuesday, May 27, 2025 at 15.00 hrs:

- A total of four **ESHG Early Career Awards** are granted for outstanding research by early career scientists presented as a spoken contribution at the conference.
- The **Isabelle Oberlé Award** is awarded yearly since 2002 for best presentation by an early career scientist on research concerning the genetics of intellectual disability.
- The Lodewijk Sandkuijl Award was instituted in 2004 to be awarded to the author of the best
  presentation at the ESHG conference within the field of complex disease genetics and statistical
  genetics.
- The **Vienna Medical Academy Award** will be awarded to the best presentation in translational genetics therapy of genetic diseases.
- The Mia Neri Award will be awarded to the best presentation in cancer research.

All winners will receive a monetary prize in the amount of EUR 600, a complimentary ESHG membership for one year as well as a free participation in next year's conference.

Talks of ECA candidates are marked with an asterisk \* in the programme overview.

Participants can find the profiles of all candidates in the virtual conference platform under the menu option presenters using the filter: Early Career Awardees/ECA

### https://cattendee.abstractsonline.com/meeting/21105

Additionally, the list can be found on the ESHG Mobile App under the menu Programme.

### ESHG Early Career Poster Award Candidates

The ESHG proposes the ESHG Early Career Poster Awards for the best posters presented by Early Career Investigators at the meeting. The two winners (one in clinical, the other in basic research) will receive a prize money of EUR 600, a complimentary ESHG membership for one year as well as a free participation in next year's conference.

The five honorable mentions receive a complimentary ESHG membership for one year.

The ESHG Scientific Programme Committee has selected a number of candidates for the ESHG Early Career Poster Awards based on the score of their submission after peer review. Candidate posters can be identified by a rosette on the board.

Participants can find the profiles of all candidates in the virtual conference platform under the menu option presenters using the filter: Early Career Awardees/ECA

https://cattendee.abstractsonline.com/meeting/21105

Additionally, the list can be found on the ESHG Mobile App under the menu Posters.





### **Hybrid Posters**

All Hybrid Posters will be presented at the in-person conference as a paper poster in the exhibition hall 4. Additionally, presenters will upload an e-poster on the virtual conference platform.

01. Cancer Genetics
02. Reproductive Genetics
03. Prenatal Genetics
04. Sensory Disorders (Eye, Ear, Pain)
05. Internal Organs and Endocrinology (Lung, Kidney, Liver, Gastrointestinal) P05.001 – P05.069
06. Skeletal, Connective Tissue, Ectodermal and Skin Disorders
07. Cardiovascular Disorders
08. Metabolic and Mitochondrial Disorders
09. Immunology and Hematopoietic System
10. Intellectual Disability
11. Neurogenetic and Psychiatric Disorders
12. Neuromuscular Disorders
13. Multiple Malformation/Anomalies Syndromes
14. Cytogenetics, Genome Variation and Architecture
15. Genomics of rare diseases
16. Diagnostic Improvements and Quality Control
17. Bioinformatics, Machine Learning and Statistical Methods
18. Large scale genomics (GWAS) and other -omics association studies
19. Genetic Epidemiology and Mendelian Randomisation
20. Pharmacogenomics and Drug Repurposing
21. Population Genetics and Evolutionary Genetics
22. Functional Genomics
23. New Technologies
24. Treatments for Genetic Disorders
25. Genetic Counselling/Services/Education
26. Ethical, Legal and Psychosocial Aspects in Genetics

### Information for Presenters of Hybrid Posters

Posters will be on display from:Saturday, May 24, (09.30 hrs) to Monday, May 26 (17.00 hrs)Poster mounting will be possible on:Saturday, May 24, from 09.30 hrs onwardsRemoval will be mandatory on:Groups A-C: Monday, May 26, 2025: 16.45 – 17.00 hrsGroup D: Monday, May 26, 2025: 17.00 – 17.05 hrs

You can find your poster board number on the ESHG 2025 mobile app or on the 2025 ESHG virtual conference platform.

Access after Monday, May 26, 17.00 hrs is not possible! Safety regulations in place for the exhibition break-down do not allow participants in the hall after this time. Please note that posters not removed until this time will be taken down by the staff of the conference centre.

They will be available for (unsupervised) pickup until Tuesday, 14.00 hrs, but will not be stored afterwards or sent to the authors after the meeting.

### Hybrid Poster Presentations

In order to enable discussion and interaction with other participants, it is mandatory for you or one of your group members to be at your poster board at the following time:

- Poster Group A: 13.00 14.00 hrs CEST on Sunday, May 25 for posters with board numbers ending with "A" (e.g. P01.001.A)
- Poster Group B: 16.00 17.00 hrs CEST on Sunday, May 25 for posters with board numbers ending with "B" (e.g. P01.001.B)
- **Poster Group C:** 13.00 14.00 hrs CEST on Monday, May 26 for posters with board numbers ending with "C" (e.g. P01.001.C)
- Poster Group D: 16.00 17.00 hrs CEST on Monday, May 26 for posters with board numbers ending with "D" (e.g. P01.001.D)

If it is not possible for you or one of your group members to be present during the above stated times, please leave a note on your poster board detailing the times when you will be present at the board.

These presentations will only take place in-person in Hall 4. There is no virtual presentation for hybrid posters.

Please note that taking pictures is allowed, unless requested otherwise by the presenter.

### **Best Hybrid Poster Presentations**

The 30 Best Hybrid Posters were selected for a short presentation (3 minutes) in three Concurrent Sessions – C08, C23 & C31 on Saturday, Sunday & Monday from 10.30 - 12.00 hrs. These sessions will take place at the Sequencing Square in the exhibition hall 4. After the presentations, participants will have the opportunity to approach the presenters of each poster for questions at the E-Poster screens next to the Sequencing Square.

### **E-Posters**

E-Posters will be available digitally on the virtual conference platform. Additionally, in-person participants can make use of the ePoster screens available in Hall 4 at the E-Poster area to browse through all E-Posters and Hybrid E-Posters that have been uploaded to the virtual conference platform. These posters have also been grouped based on the abstract submission topics (please refer to the Hybrid Posters information for a detailed overview of all topics).

### Abstracts

Abstracts presented at the ESHG 2025 conference can be found on the following platforms:

- ESHG Virtual Conference Platform
- ESHG Mobile App
- online programme on website

Poster Hall Floor Plan >>> go to pages 22-23

#### **PROGRAMME BUSINESS AND ANCILLARY MEETINGS** 18

As per 04.05.2025

### Friday, May 23, 2025

Time	Meeting	Room	
08.30 – 12.30 hrs	ESHG Executive Board Meeting	. Suite 7	closed
13.00 – 18.00 hrs	ESHG Board Meeting	. Amber 4	closed

### Saturday, May 24, 2025

Time	Meeting	Room
09.00 – 10.00 hrs	ESHG Social Media Committee	. Amber 4 closed
09.00 – 13.30 hrs	ESHG PEC Meeting	. Suite 5 closed
11.00 – 13.30 hrs	ESHG EuroGentest Committee Meeting	. Suite 7 closed
14.00 – 17.00 hrs	MDPI Editorial Board Meeting	. Suite 5 closed
17.30 – 18.15 hrs	ESHG-Y Committee	. Suite 7 closed

### Sunday, May 25, 2025

Time	Meeting	Room
08.30 – 09.30 hrs	Springer Nature – Editorial Board Meeting Journal	
	of Community Genetics	. Suite 5 closed
09.00 – 11.00 hrs	European Network of Genetic Nurses and Counsellors	
	Meeting organized by the GNGC branch, EBMG	
09.30 – 11.30 hrs	ESHG Strategic Committee Meeting	
12.00 – 14.00 hrs	National Human Genetics Societies Meeting	
12.15 – 13.15 hrs	ESHG Podcast – Editors' Meeting	
13.30 – 15.15 hrs	GenIDA Scientific Advisory Board Meeting	
14.00 – 15.00 hrs	EJHG Editorial Board Meeting	
14.30 – 17.00 hrs	UEMS – Section of Medical Genetics	
15.45 – 16.45 hrs	ERN-ITHACA ExCom Meeting	
16.00 – 18.00 hrs	ErCLG board meeting	
16.00 – 17.00 hrs	ESHG/ASHG Leadership Meeting	
16.15 – 17.15 hrs	TAGC Networking Event	. Suite 8 closed
17.00 – 18.30 hrs	EMQN workshop: 25 years of HGVS:	
	Updates and advancements in ensuring accurate	
	description of sequence variants	•
18.00 – 20.00 hrs	1+MG and AllofUS collaboration	
18.45 – 19.45 hrs	ERN-ITHACA Intermediary Board Meeting	. Amber 4 closed

### Monday, May 26, 2025

Time	Meeting	Room
08.30 – 10.30 hrs	ESHG Education Committee Meeting	. Suite 5 closed
11.00 – 13.00 hrs	Preconception carrier screening: what to include and why?.	. Amber 4 closed
12.15 – 13.15 hrs	ESHG General Assembly	. Brown 3 open to
		ESHG members
13.15 – 14.15 hrs	ESHG ERN Coordinators	. Suite 5 closed
13.30 – 15.30 hrs	EBMG General Assembly	. Amber 4 closed
15.00 – 16.30 hrs	IFHGS Executive Board Meeting	. Suite 5 closed
16.00 – 17.30 hrs	EBMG Branch of Medical Geneticists	. Suite 7 closed
17.00 – 18.30 hrs	ICHG SPC Meeting	. Suite 5 closed
Tuesday, May 2	7, 2025	

### Tuesday, May 27, 2025

Time	Meeting	Room
12.30 – 13.30 hrs	ESHG SPC Meeting	Amber 4 closed

#### Disclaimer

Ancillary and satellite meetings shall not state or imply endorsement of, or support by the ESHG of the event, organiser, products or services presented in any verbal statements or printed/electronic media before, after and during the presentations.





Overview of Corporate Sessions (Amber rooms) and Presentations (Sequencing Square, Exhibition Hall 4) in order of date & time.

Saturday 24 May10.00 - 11.15 hrsLevel 2enGenome - Transforming variant assessment with Al: eVai in Diagnostics and VarChat on UCSCAmber 2300Saturday 24 May12.15 - 13.30 hrsLevel 2Blueprint Genetics - Empowering Precision Medicine: The power of WES, mitochondrial analysis, WGS and screeningAmber 5+6216MGI TECH - Advancing Clinical and Population Genomics with MGI's DNBSEQ and Multi-Omics SolutionsAmber 7+8310QIAGEN - Redefining Tumor Profiling: An Innovative, Non-Invasive Approach to cfDNA Biomarker Integration from Plasma and Urine Using Advanced BioinformaticsAmber 2330SOPHiA GENETICS - Applied innovation in genomics: Enhanced exome and hereditary cancer end-to-end solutionsAmber 3460varvis* - Read long and prosper: Efficient long-read data analysis with the varvis* osftware.Amber 1220Sunday 25 May08.30 - 10.00 hrsLevel 211Illumina - Innovations driving the Multiomics revolutionAmber 5+6430Watchmaker Genomics - Sequenomics: Automated workflows for genomic variant detectionAmber 7+8530Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders: Novel Approach Tackling Complex Variants in challenging Genes with Nanopore SequencingAmber 2376Covaris - Comprehensive Sample Preparation for Accurate and Reliable DataAmber 3144Medicover Genetics - Scalable Technology Infinited Workflows to Maximize Efficiency automated workflows to Dimized Workflows to Maximize KfinciencyAmber 1486Olink Proteomics - From Pilot to Breakthroughs: Insights from the	Company	loom Sta	nd #
eVai in Diagnostics and VarChat on UCSCAmber 2300Saturday 24 May12.15 - 13.30 hrsLevel 2Blueprint Genetics - Empowering Precision Medicine: The power of WES, mitochondrial analysis, WGS and screeningAmber 5+6216MGI TECH - Advancing Clinical and Population Genomics with MGI's DNBSEQ and Multi-Omics SolutionsAmber 7+8310QIAGEN - Redefining Tumor Profiling: An Innovative, Non-Invasive Approach to cfDNA Biomarker Integration from Plasma and Urine Using Advanced BioinformaticsAmber 2330SOPHiA GENETICS - Applied innovation in genomics: Enhanced exome and hereditary cancer end-to-end solutionsAmber 3460varvis® - Read long and prosper: Efficient long-read data analysis with the varvis® software.Amber 1220Sunday 25 May08.30 - 10.00 hrsLevel 211Illumina - Innovations driving the Multiomics revolutionAmber 5+6430Watchmaker Genomics - Sequenomics: Maximizing Data Quality, Minimizing Process Complexity in NGSAmber 3288Sunday 25 May12.00 - 13.00 hrsLevel 2Agilent Technologies - Enhancing rare disease research through automated workflows for genomic variant detectionAmber 7+8530Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders: Novel Approach Tackling Complex Variants in challenging Genes with Nanopore SequencingAmber 3144Medicover Genetics - Scalable Technology Platform for Genetic Testing: CE-marked and IVDR Solutions with Optimized Workflows to Maximize EfficiencyAmber 1486Olink Proteomics - From Pilot to Breakthroughs: Insights from the UK	Saturday 24 May 10.00 - 11.15 hrs Level 2		
Blueprint Genetics - Empowering Precision Medicine:       Amber 5+6       216         MGI TECH - Advancing Clinical and Population Genomics       Amber 5+6       216         MGI TECH - Advancing Clinical and Population Genomics       Amber 7+8       310         QIAGEN - Redefining Tumor Profiling: An Innovative, Non-Invasive Approach to cfDNA Biomarker Integration from Plasma and Urine Using Advanced Bioinformatics       Amber 2       330         SOPHiA GENETICS - Applied innovation in genomics:       Enhanced exome and hereditary cancer end-to-end solutions       Amber 3       460         varvis <sup>®</sup> - Read long and prosper: Efficient long-read data analysis with the varvis <sup>®</sup> software.       Amber 1       220         Sunday 25 May       08.30 - 10.00 hrs       Level 2         Illumina - Innovations driving the Multiomics revolution       Amber 5+6       430         Watchmaker Genomics - Sequenomics: Maximizing Data Quality, Minimizing Process Complexity in NGS       Amber 3       288         Sunday 25 May       12.00 - 13.00 hrs       Level 2       Amber 7+8       530         Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders: Novel Approach Tackling Complex Variants in challenging Genes       Amber 3       144         Medicover Genetics - Scalable Technology Platform for Genetic Testing: CE-marked and IVDR Solutions with Optimized Workflows to Maximize Efficiency Amber 3       144		Amber 2	300
The power of WES, mitochondrial analysis, WGS and screening			
MGI TECH - Advancing Clinical and Population Genomics       Amber 7+8       310         QIAGEN - Redefining Tumor Profiling: An Innovative, Non-Invasive Approach to cfDNA Biomarker Integration from Plasma and Urine Using Advanced Bioinformatics       Amber 2       330         SOPHiA GENETICS - Applied innovation in genomics: Enhanced exome and hereditary cancer end-to-end solutions       Amber 3       460         varvis® - Read long and prosper: Efficient long-read data analysis with the varvis® software.       Amber 1       220         Sunday 25 May       08.30 - 10.00 hrs       Level 2       Illumina - Innovations driving the Multiomics revolution       Amber 5+6       430         Watchmaker Genomics - Sequenomics: Maximizing Data Quality, Minimizing Process Complexity in NGS       Amber 1       288         Sunday 25 May       12.00 - 13.00 hrs       Level 2       Illumina - Innovations for genomic variant detection       Amber 7+8       530         Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders: Novel Approach Tackling Complex Variants in challenging Genes with Nanopore Sequencing       Amber 2       376         Covaris - Comprehensive Sample Preparation for Accurate and Reliable Data       Amber 3       144         Medicover Genetics - Scalable Technology Platform for Genetic Testing: CE-marked and IVDR Solutions with Optimized Workflows to Maximize Efficiency       Amber 1       486	Blueprint Genetics - Empowering Precision Medicine:		
QIAGEN - Redefining Tumor Profiling: An Innovative, Non-Invasive Approach to cfDNA Biomarker Integration from Plasma and Urine Using Advanced BioinformaticsAmber 2330SOPHIA GENETICS - Applied innovation in genomics: Enhanced exome and hereditary cancer end-to-end solutionsAmber 3460varvis® - Read long and prosper: Efficient long-read data analysis with the varvis® software.Amber 1220Sunday 25 May08.30 - 10.00 hrsLevel 2Illumina - Innovations driving the Multiomics revolutionAmber 5+6430Watchmaker Genomics - Sequenomics: Maximizing Data Quality, Minimizing Process Complexity in NGSAmber 3288Sunday 25 May12.00 - 13.00 hrsLevel 2Agilent Technologies - Enhancing rare disease research through automated workflows for genomic variant detectionAmber 7+8530Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders: Novel Approach Tackling Complex Variants in challenging Genes with Nanopore SequencingAmber 3144Medicover Genetics - Scalable Technology Platform for Genetic Testing: CE-marked and IVDR Solutions with Optimized Workflows to Maximize Efficiency CE-marked and IVDR Solutions with Optimized Workflows to Maximize EfficiencyAmber 1486	<b>MGI TECH</b> - Advancing Clinical and Population Genomics		
SOPHiA GENETICS - Applied innovation in genomics: Enhanced exome and hereditary cancer end-to-end solutions Amber 3460varvis® - Read long and prosper: Efficient long-read data analysis with the varvis® software			310
Enhanced exome and hereditary cancer end-to-end solutionsAmber 3460varvis® - Read long and prosper: Efficient long-read data analysis with the varvis® software.Amber 1220Sunday 25 May08.30 - 10.00 hrsLevel 2Illumina - Innovations driving the Multiomics revolutionAmber 5+6430Watchmaker Genomics - Sequenomics: Maximizing Data Quality, Minimizing Process Complexity in NGSAmber 3288Sunday 25 May12.00 - 13.00 hrsLevel 2Agilent Technologies - Enhancing rare disease research through automated workflows for genomic variant detectionAmber 7+8530Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders: Novel Approach Tackling Complex Variants in challenging Genes with Nanopore SequencingAmber 2376Covaris - Comprehensive Sample Preparation for Accurate and Reliable DataAmber 3144Medicover Genetics - Scalable Technology Platform for Genetic Testing: CE-marked and IVDR Solutions with Optimized Workflows to Maximize EfficiencyAmber 1486Olink Proteomics - From Pilot to Breakthroughs: Insights from the UK Biobank200		Amber 2	330
with the varvis® software.Amber 1220Sunday 25 May08.30 - 10.00 hrsLevel 2Illumina - Innovations driving the Multiomics revolutionAmber 5+6430Watchmaker Genomics - Sequenomics: Maximizing Data Quality, Minimizing Process Complexity in NGSAmber 5+6430Sunday 25 May12.00 - 13.00 hrsLevel 2Agilent Technologies - Enhancing rare disease research through automated workflows for genomic variant detectionAmber 7+8530Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders: Novel Approach Tackling Complex Variants in challenging Genes with Nanopore SequencingAmber 2376Covaris - Comprehensive Sample Preparation for Accurate and Reliable DataAmber 3144Medicover Genetics - Scalable Technology Platform for Genetic Testing: CE-marked and IVDR Solutions with Optimized Workflows to Maximize EfficiencyAmber 1486Olink Proteomics - From Pilot to Breakthroughs: Insights from the UK BiobankSinte Proteomice - From Pilot to Breakthroughs: Insights from the UK BiobankSinte Proteomice - Sinte Pilot to Breakthroughs: Insights from the UK BiobankSinte Pilot to Breakthroughs: Insights from the UK Biobank	Enhanced exome and hereditary cancer end-to-end solutions	Amber 3	460
Illumina - Innovations driving the Multiomics revolutionAmber 5+6430Watchmaker Genomics - Sequenomics: Maximizing Data Quality, Minimizing Process Complexity in NGSAmber 3288Sunday 25 May12.00 - 13.00 hrsLevel 2Agilent Technologies - Enhancing rare disease research through automated workflows for genomic variant detectionAmber 7+8530Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders: Novel Approach Tackling Complex Variants in challenging Genes with Nanopore SequencingAmber 2376Covaris - Comprehensive Sample Preparation for Accurate and Reliable DataAmber 3144Medicover Genetics - Scalable Technology Platform for Genetic Testing: CE-marked and IVDR Solutions with Optimized Workflows to Maximize EfficiencyAmber 1486Olink Proteomics - From Pilot to Breakthroughs: Insights from the UK BiobankAmber 1486	with the varvis <sup>®</sup> software	Amber 1	220
Watchmaker Genomics - Sequenomics: Maximizing Data Quality, Minimizing Process Complexity in NGS       Amber 3       288         Sunday 25 May       12.00 - 13.00 hrs       Level 2         Agilent Technologies - Enhancing rare disease research through automated workflows for genomic variant detection       Amber 7+8       530         Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders: Novel Approach Tackling Complex Variants in challenging Genes with Nanopore Sequencing       Amber 2       376         Covaris - Comprehensive Sample Preparation for Accurate and Reliable Data       Amber 3       144         Medicover Genetics - Scalable Technology Platform for Genetic Testing: CE-marked and IVDR Solutions with Optimized Workflows to Maximize Efficiency       Amber 1       486         Olink Proteomics - From Pilot to Breakthroughs: Insights from the UK Biobank       Amber 1       486			
Minimizing Process Complexity in NGS       Amber 3       288         Sunday 25 May       12.00 - 13.00 hrs       Level 2         Agilent Technologies - Enhancing rare disease research through automated workflows for genomic variant detection       Amber 7+8       530         Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders: Novel Approach Tackling Complex Variants in challenging Genes with Nanopore Sequencing       Amber 2       376         Covaris - Comprehensive Sample Preparation for Accurate and Reliable Data       Amber 3       144         Medicover Genetics - Scalable Technology Platform for Genetic Testing: CE-marked and IVDR Solutions with Optimized Workflows to Maximize Efficiency       Amber 1       486         Olink Proteomics - From Pilot to Breakthroughs: Insights from the UK Biobank       Amber 1       486		Amber 5+6	430
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Asuragen, a Bio-Techne brand - Precision Screening for Inherited Disorders:       Novel Approach Tackling Complex Variants in challenging Genes       376         Novel Approach Tackling Complex Variants in challenging Genes       Amber 2       376         Covaris - Comprehensive Sample Preparation for Accurate and Reliable Data       Amber 3       144         Medicover Genetics - Scalable Technology Platform for Genetic Testing:       Technarked and IVDR Solutions with Optimized Workflows to Maximize Efficiency       Amber 1       486         Olink Proteomics - From Pilot to Breakthroughs: Insights from the UK Biobank       Amber 1       486		Ample or 7 - 0	520
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Medicover Genetics - Scalable Technology Platform for Genetic Testing:         CE-marked and IVDR Solutions with Optimized Workflows to Maximize Efficiency       Amber 1       486         Olink Proteomics - From Pilot to Breakthroughs: Insights from the UK Biobank       486		Amber 2	376
Olink Proteomics - From Pilot to Breakthroughs: Insights from the UK Biobank	Medicover Genetics - Scalable Technology Platform for Genetic Testing:	Amber 3	144
Pharma Proteomics Project and what's next for proteomics         Amber 5+6         388	<b>Olink Proteomics</b> - From Pilot to Breakthroughs: Insights from the UK Biobank	Amber 1	486
	Pharma Proteomics Project and what's next for proteomics	Amber 5+6	388
Sunday 25 May 12.15 - 12.45 hrs Level 2			
Stilla Technologies - Monitoring CAR-T Cell Therapy in Mantle Cell Lymphoma Using cfDNA and dPCR: Innovative Approaches.         Sequencing Square         Sequencing 548			548
Sunday 25 May 13.00 - 13.30 hrs Level 2	Sunday 25 May 13.00 - 13.30 hrs Level 2		
Thermo Fisher Scientific - Genetic Innovation: Exploring IVDR-Compliant Sanger         Sequencing           Sequencing for Enhanced Diagnostics         Square         520			520
Sunday 25 May 14.15 - 15.45 hrs Level 2	Sunday 25 May 14.15 - 15.45 hrs Level 2		
<b>10x Genomics</b> - Cutting-Edge Solutions for Genomic Insights       Amber 3       202 <b>Element Biosciences</b> - Unlock high dimensional biology using next		Amber 3	202
next-generation-sequencing	next-generation-sequencing	Amber 2	392
PacBio - It starts with HiFi.   Amber 5+6   454		Amber 5+6	454
Roche - Enabling rare disease research with rapid workflows by SBX technology and the AVENIO Edge automated KAPA HyperExome V2 solution		Amber 1	490

### INDUSTRY CORPORATE SESSIONS & PRESENTATIONS 21

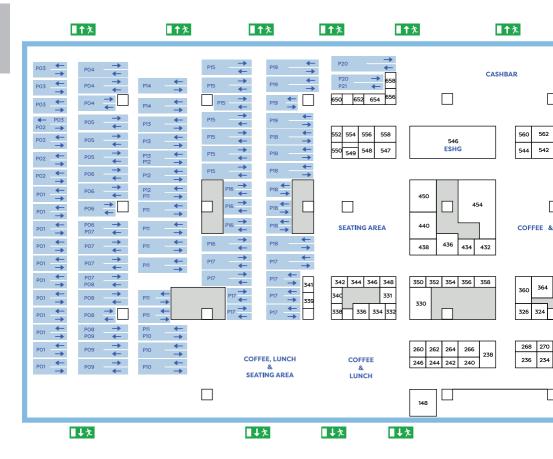
Overview of Corporate Sessions (Amber rooms) and Presentations (Sequencing Square, Exhibition Hall 4) in order of date & time.

Company	Room	Stand #
Monday 26 May 12.00 - 13.00 hrs Level 2		
Oxford Nanopore Technologies - Shaping the landscape of rare disease research		
in Europe	. Amber	7+8 400
Parse Biosciences - Redefining Single Cell RNA Sequencing Limits		
with Evercode <sup><math>m</math></sup> Combinatorial Barcoding $\ldots$ $\ldots$ $\ldots$ $\ldots$ $\ldots$ $\ldots$	. Amber	2 260
<b>Standard BioTools</b> - High-plex proteomics for fully informed genomics analysis.	. Amber	3 438
Twist Bioscience - Unleash the Power of Your Samples:Next-Level Sequencing		
with Twist Bioscience	. Amber	5+6 472
Monday 26 May 12.15 - 12.45 hrs Level 2		
<b>Thermo Fisher Scientific</b> - Innovative Breakthroughs in Pharmacogenetic Testing: Pioneering the Future of Precision Medicine		5
	. Square	520
Monday 26 May 14.15 - 15.45 hrs Level 2		
Genomics England - Unlocking discovery via one of the world's largest rare disease and cancer whole genome sequencing datasets           Illumina - Breaking new grounds in Genomics	. Amber	Not 2 exhibiting 7+8 430

The sponsors of the Corporate Sessions and Presentations planned within the main conference programme are approved as reputable and relevant by the Scientific Programme Committee. However, the detailed content of the presentations is proposed by the sponsors and under their responsibility. Neither the ESHG, nor the Conference organisers have endorsed the content in any way.



### 22 INDUSTRY FLOOR PLAN - EXHIBITION & POSTER HALL

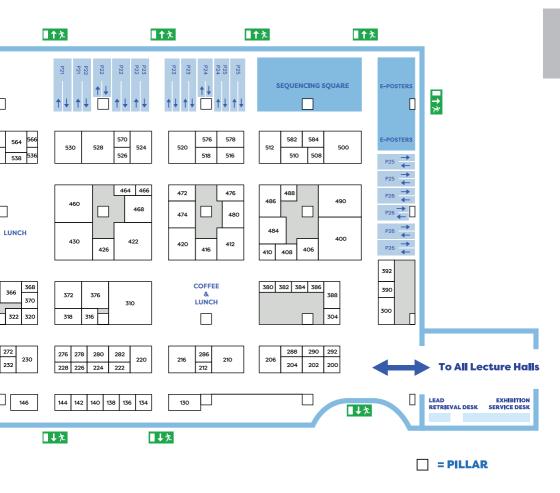




- **01. Cancer Genetics**
- 02. Reproductive Genetics
- 03. Prenatal Genetics
- 04. Sensory Disorders (Eye, Ear, Pain)
- 05. Internal Organs and Endocrinology
- (Lung, Kidney, Liver, Gastrointestinal) 06. Skeletal, Connective Tissue,
  - Ectodermal and Skin Disorders

- 07. Cardiovascular Disorders
- 08. Metabolic and Mitochondrial Disorders
- 09. Immunology and Hematopoietic System
- 10. Intellectual Disability
- 11. Neurogenetic and Psychiatric Disorders
- 12. Neuromuscular Disorders
- 13. Multiple Malformation/Anomalies Syndromes

### INDUSTRY FLOOR PLAN - EXHIBITION & POSTER HALL



- 14. Cytogenetics, Genome Variation and Architecture
- 15. Genomics of rare diseases

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- 16. Diagnostic Improvements and Quality Control
- 17. Bioinformatics, Machine Learning and Statistical Methods
- 18. Large scale genomics (GWAS) and other-omics association studies
- 19. Genetic Epidemiology and Mendelian Randomisation
- 20. Pharmacogenomics and Drug Repurposing
- 21. Population Genetics and Evolutionary Genetics
- 22. Functional Genomics
- 23. New Technologies
- 24. Treatments for Genetic Disorders
- 25. Genetic Counselling/Services/Education
- 26. Ethical, Legal and Psychosocial Aspects in Genetics

List correct as per date of printing - Exhibition floor plan available in the Registration Area.

10x Genomics	202	Element Biosciences	392
		Empire Genomics	136
3billion	390	EMQN CIC	358
		enGenome	300
4bases	560	ESHG - European Society of Human Genetics	546
		Euformatics	380
Abomics Oy	354	Eurofins Genomics	364
ADS BIOTEC	142	Eurofins Genoma Group	584
Agena Bioscience	270	EUROPEAN REFERENCE NETWORKS	550
Agilent Technologies	530		
AIBWS ODV	650	FamGenix	508
Alida Biosciences	138	Franklin by Genoox	518
American Society of Human Genetics	552	Fulgent Genetics	324
APIS Assay Technologies	286		
Applied Spectral Imaging	656	Galatek	440
Arcensus	146	GeneMind Biosciences	500
AstraFormedic Gruppo A. De Mori,		GENERI BIOTECH	348
co-exhibitor of LGC Clinical Diagnostics	564	Genes2Me	524
Asuragen, a Bio-Techne brand	376	Genetek Biopharma	292
		GENEWIZ By Azenta Life Sciences	549
bio.logis digital health	466	Geneyx	578
Bio Molecular Systems	360	Genome Diagnostics Nijmegen Maastricht	318
Biomarker Technologies (BMK)	326	Genomenon	200
Bionano	356	GenomeUp	488
BioView	228	GenomeWeb	368
Blueprint Genetics	216	Genomize	542
Breda Genetics	290	GenQA	412
Burning Rock Dx	234	GILSON	547
5		Golden Helix	510
CeGaT	372		
Celemics	230	HAMILTON BONADUZ	512
CENTOGENE	528	Hangzhou Allsheng Instruments	238
Centro Nacional de Análisis Genómico (CNAG)	382	Hangzhou Diagens Biotechnology	140
CHAMP1 Foundation - Europe	652	Health in Code	210
Clinisys	538		
COCOON BIOSCIENCE	332	iECURE	232
College of American Pathologists	272	IGATech	266
Copan	526	Illumina	430
Covaris	144	INOREVIA	262
CyberGene	566	Integrated DNA Technologies	468
		Isohelix	316
Devyser	436		
Diatech Pharmacogenetics	450	JSI medical systems	206
DNA Genotek	406		
DNA LINK	331		
Dovetail Genomics	264		

# INDUSTRY LIST OF EXHIBITORS 25

Labcorp	341	QIAGEN	330
LABGENETICS	320	Quantabio	434
LaCAR MDx Technologies	432		
LGC Clinical Diagnostics	564	RESNOVA, co-exhibitor of Biomolecular Systems	360
LIFE & BRAIN	422	Revvity	130
Longlight Technology	576	Roche	490
Lucid Genomics	558		
		Salus BioMed	212
Macrogen Europe	370	Saphetor	426
Magtivio	342	SCC Soft Computer	536
MDPI - Academic Open Access Publishing since		SegOIA, co-exhibitor of OVHcloud	554
1996	244	SegOne	474
Medicover Genetics	486	Seguins	346
Meridian Bioscience - Life Science Division	280	SOPHIA GENETICS	460
MetaSystems	350	SPRINGER NATURE	544
MGITÉCH	310	SPT Labtech	240
MGZ Munich	410	Standard BioTools	438
MRC Holland	416	Steribar Systems	336
		Stilla Technologies	548
Nanodigmbio	222	SYNLAB	204
Negedia	464	Sysmex Europe	582
New England Biolabs	480		
Nexco Analytics	242	Takara Bio Europe	352
NEXT LAB ITALY	339	Tecan Italia	366
NEXUS / GEPADO - software solutions for geneti	CS	TECHNIDATA	334
278		TECHNOGENETICS	570
Nightingale Health	134	Thermo Fisher Scientific	520
NimaGen	516	TRUPCR Europe	322
Nostos Genomics	304	Twist Bioscience	472
Novogene	276		
Nutanix, co-exhibitor of OVHcloud	554	UNIAMO Federazione Italiana Malattie Rare	654
		UCSC Genome Browser	226
Olink Proteomics	388		
Omega Bio-tek	282	varvis®	220
OmicsDiscoveries	338	Vazyme	224
OMICS EMPOWER	236	Velsera	562
Opentrons	384	Veritas Genetics	484
ORPHANET - INSERM US14	340	Volta Labs	658
OVHcloud	554		
Oxford Nanopore Technologies	400	Watchmaker Genomics	288
		Wisepress Medical Bookshop	148
PacBio	454		
Paragon Genomics	268	Yourgene Health	408
Parse Biosciences	260		
PCR Biosystems	556	Zymo Research Europe	476
PhenoTips	386		
PhiTech Bioinformatics	246		
PLATOMICS	344		
Promega Corporation	420		

### Exhibition & Poster Area – Hall 4 – Dates & Opening Hours (CEST)

Saturday, May 24	09.30 – 18.30 hrs
Sunday, May 25	09.30 – 17.00 hrs
Monday, May 26	09.30 – 17.00 hrs
Tuesday, May 27	CLOSED

### **Exhibitor Information**

Exhibitor Information, as well as the Exhibitor Index of Products and Services may be found in the ESHG 2025 Conference App. Download the App, for iOS or Android, from iTunes App Store and Google Play Store.

### **Corporate Session & Presentation Programmes**

In the ESHG 2025 Conference App you will find the Corporate Session\* and Presentation\* Programmes. For a printed overview of all Corporate Sessions and Presentations, including session titles, please check the Conference Bag counter immediately after the Registration Area.

\*Corporate Sessions will be held in the Amber rooms, Corporate Presentations at the Sequencing Square in the Exhibition Hall.

### Floor Plan – Exhibition & Poster Topics

You will find the Exhibition & Poster Topics plan in the Conference Bag counter immediately after the Registration Area.

### Posters – Mounting, Viewing & Removal Schedules

Poster presentations will be held in the Exhibition & Poster Hall from 24 – 26 May.

Poster mounting, viewing and removal times are:

Saturday, May 24	09.30 – 18.30 hrs	Poster mounting / viewing
Sunday, May 25	09.30 – 17.00 hrs	Poster viewing
Monday, May 26	09.30 – 16.45 hrs	Poster viewing
	16.45 – 17.05 hrs	Poster removal

Posters not removed by 17.05 hrs on Monday 26 May will be taken down and will not be stored or sent to authors after the meeting.

### INDUSTRY INFORMATION EXHIBITION & POSTER AREA 27

### Coffee Breaks, Cash Bar, Lunch

Official coffee breaks, as per the conference programme, will be held in the Exhibition & Poster Hall on Saturday, Sunday and Monday. Also outside the official coffee break times there will be free coffee and tea in the Exhibition Hall.

Coffee breaks & lunch on Tuesday: Coffee Breaks: Balcony (level 1), Bar Milano & Cube Lounge (level 2) Lunch: South Hall (level 0)

The Cash Bar in the Exhibition Hall is open during exhibition opening hours. The menu includes sandwiches, salads, selection of warm dishes and drinks. The menu is available at the Cash Bar. Payment by card and cash.

Pre-ordered lunch bags will be available during lunch breaks at the coffee break areas. Lunch bags cannot be ordered on-site.

### Lead Retrieval System used by Companies

Many companies will be using a so-called Lead Retrieval System on their stands and at the entrance to their Corporate Session.

Note the following please:

- Companies using the device must ask permission to scan the QR code on your badge. Refusal to have your badge scanned does not entitle a company to deny you access to their Corporate Session or to join an activity at their stand.
- The QR code on your badge gives the relevant company access to your contact details as follows:
  - o name and institution/organisation
  - o e-mail address
  - o country

Your contact details will only be shared with the relevant companies in case you opted for this during the registration process and/or if you agreed on-site to your badge being scanned.

Thank you for your understanding and cooperation.



### 28 INFORMATION GENERAL INFORMATION

### **Registration and Opening Hours**

### **Opening Hours Registration**

Friday, May 23	14.00 – 18.00 hrs CEST
Saturday, May 24	07.30 – 20.15 hrs CEST
Sunday, May 25	08.00 – 19.00 hrs CEST
Monday, May 26	08.00 – 19.00 hrs CEST
Tuesday, May 27	08.30 - 14.00 hrs CEST

#### **Opening Hours Speaker Service Centre**

14.00 – 18.00 hrs CEST
07.30 - 20.15 hrs CEST
08.00 - 19.00 hrs CEST
08.00 - 19.00 hrs CEST
08.30 – 15.00 hrs CEST

### **Opening Hours Exhibition and Poster Area**

 Friday, May 23
 CLOSED!

 Saturday, May 24
 09.30 – 18.30 hrs CEST

 Sunday, May 25
 09.30 – 17.00 hrs CEST

 Monday, May 26
 09.30 – 17.00 hrs CEST

 Tuesday, May 27
 CLOSED!

### Cloakroom

Saturday, May 24	07.30 – 22.00 hrs CEST
Sunday, May 25	08.00 – 20.30 hrs CEST
Monday, May 26	08.00 – 02.30 hrs CEST
Tuesday, May 27	08.30 – 17.00 hrs CEST

### **Childcare Service**

Saturday, May 24	08.00 - 20.30 hrs CEST
Sunday, May 25	08.00 - 19.15 hrs CEST
Monday, May 26	08.00 - 19.15 hrs CEST
Tuesday, May 27	08.30 - 16.30 hrs CEST

Please note that the childcare service must have been booked in advance and can only be accommodated upon availability on-site.



Want to know more

### INFORMATION GENERAL INFORMATION 29



#### Late programme changes

All contents are up-to-date as per date of printing (05.05.2025). For the most up to date complete scientific programme please consult the programme planner, virtual conference platform or ESHG mobile app.

#### Language

The official language of the conference is English (no simultaneous translation available).

#### **Smoking Policy**

The ESHG 2025 is officially a "No-smoking-Conference". Note that smoking is banned in public buildings and private businesses – including restaurants, pubs, shops, public transport, entertainment venues and workplaces.

### **Social Media Guidelines**

Please see our full policy on our website. https://2025.eshg.org/myconference/mediaonlinepolicy/ The ESHG supports the use of social media around the European Human Genetics Conference to network with your colleagues and friends attending the meeting. Please do however respect the ESHG social media guidelines.

The views and opinions posted on ESHG's social media do not necessarily reflect the views, opinions, or policies of the ESHG, its Board or membership. The ESHG reserves the right to remove comments it deems to be inappropriate.

### **Sustainability**

The ESHG Conference is committed to host an event that bears in mind the responsible use of resources, our environment and the use of sustainable conference materials. Hence, we would like to encourage all of our participants to keep these aspect in mind when attending the in-person meeting.

### Milan – General Information

#### **Congress Venue**

Allianz MiCo, Milano Convention Centre Piazzale Carlo Magno 20149 Milan Italy https://www.micomilano.it/en



ore Information

For information about the city of Milan visit the 2025 conference website: https://2025.eshg.org/myconference/location-venue/

### **Opening Networking Mixer**

#### Saturday, May 24, 2025, 20.00 - 22.00 hrs CEST – Allianz MiCo (Balcony terrace South Hall & Balcony, level 1)

Network with your colleagues at this mixer on Saturday evening. Drinks and small snacks will be offered. *Admission is free of charge, however admission is only possible for in-person registered participants.* 

Please make sure that you have your badge with you for identification and access to the event.

Dress code: casual

### Young Human Geneticist Networking Event (at own expense)

Sunday, May 25, 2025, 20.00 hrs CEST – Location: Spoon Restaurant & Lounge (individual arrival)

https://www.spoonmilano.it/en/home-en/ Address: Viale Bligny 39, 20136 Milan

Dear young colleagues,

"Apericena" is a word meaning a fusion of aperitif and dinner and it is very popular among young Italians as it is a promise for good quality (and quantity!) buffet food and a chilled atmosphere.

So, as a special highlight for this year's ESHG in Milan, the ESHG Young Geneticists Committee (ESHG-Y) would like to invite you to our networking dinner event to exchange ideas and get to know your peers from other countries.

Please note that there are only 200 spots available, so make sure you are a part of this premiere networking exclusively for us young (a.k.a. in training or within 4 years from training completion) human geneticists. We look forward to your participation!

Dress code: casual Costs: EUR 30

A welcome drink, finger food, a cocktail, two main dishes and desserts are included in the price. *No sponsoring is used to fund any part of the evening.* 

### ESHG Networking Evening (at own expense)

### Monday, May 26, 2025, 19.00 hrs CEST - Location: South Hall, Allianz MiCo

The networking evening is a great opportunity to meet with friends and colleagues from around the world in a relaxed atmosphere, enjoying the unmatched charm and fascination of Milan. Those who have shared this evening with us in previous years know, one would not want to miss it!

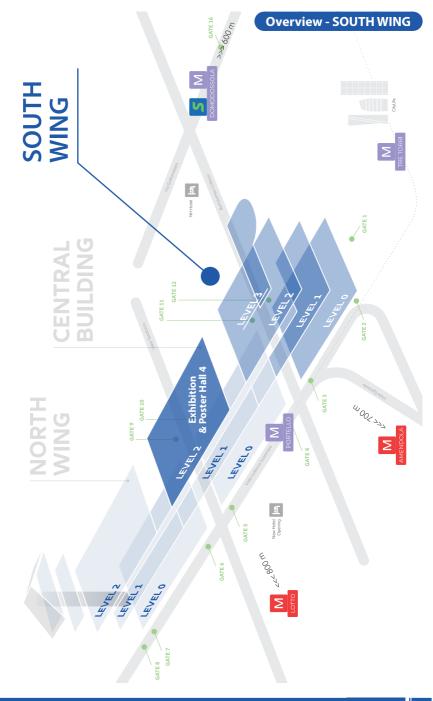
- Ticket: EUR 70.-
- Students: EUR 50.-

Dinner & 3 drinks are included in the price. Dress code: casual

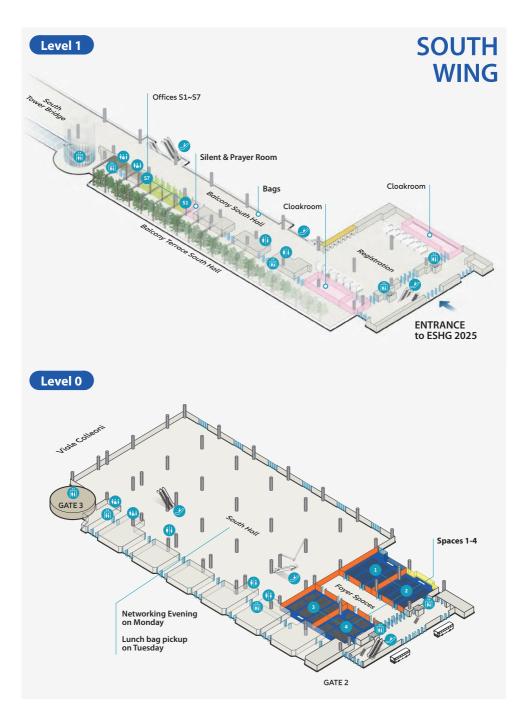
No sponsoring is used to fund any part of the evening events.



## INFORMATION GENERAL FLOORPLAN

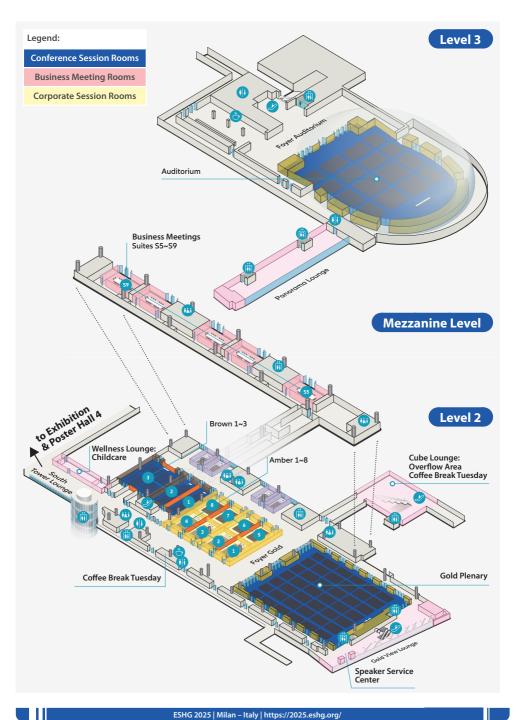


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## INFORMATION GENERAL FLOORPLAN



### **34** INFORMATION ACKNOWLEDGEMENTS

The European Human Genetics Conference gratefully acknowledges the support of the following companies.

For an overview of exhibiting companies, please see pages 24-25.

10x Genomics Agilent Technologies Arcensus Asuragen, a Bio-Techne brand Blueprint Genetics Covaris Element Biosciences enGenome Illumina Medicover Genetics

- MGI TECH MRC Holland Nabsys Olink, part of Thermo Fisher Scientific Oxford Nanopore Technologies PacBio Parse Biosciences Promega QIAGEN
- Roche Saphetor SOPHiA GENETICS Standard BioTools Stilla Technologies Thermo Fisher Scientific Twist Bioscience varvis<sup>®</sup> Velsera Watchmaker Genomics



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