Swiss Society of Medical Genetics

Register now

Main program

SSMG Annual Meeting 2025

WITH YOUNG INVESTIGATOR DAY

April 10th and 11th, 2025

University Hospital of Zurich, Lecture Hall East B10

Conference Committee

Anita Rauch
University of Zurich

Björn Kleijkers
University of Zurich

Ruxandra Bachmann-Gagescu
University of Zurich

Keynote Speakers 2025

Malte Spielmann
University Hospital Schleswig-Holstein
Beyond the genome: human genetics in the post-genomic era

Anne Goriely
University of Oxford
New mutations, selfish testes and human disease

Olivier Devuyst
University of Zurich
From rare to common: How the genetic architecture of kidney disease matters

Paranchai Boonsawat
University of Zurich
Into the unknown: long reads at the end of the odyssey

CREDIT POINTS

SGMG
Business meeting of the SSMG: 2 credits
Scientific programme SSMG meeting: 8 credits
Swiss Dysmorphology meeting: 3 credits

FAMH
Scientific programme SSMG meeting: 8 credits

REGISTRATION

Medworld AG
Sennweidstrasse 46
6312 Steinhausen

Organizers: SSMG
Björn Kleijkers | bjoern.kleijkers@medgen.uzh.ch

https://sgmg.ch/de/meeting-2025

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Thursday, April 10th, 2025 – SSMG Business Meeting & General Assembly

Chairpersons: Isabel Filges, Basel & Thierry Nouspikel, Geneva

From 08:30	**Registration open**
09:45 – 12:00	**Business Meeting of the Swiss Society of Medical Genetics**
12:00 – 13:30	*Lunch, Poster and Exhibition Viewing*
13:30 – 14:00	**Satellite Symposium organized by varvis®** **Exome diagnostics and beyond** **How standardized filtering strategies accelerate exome diagnostics** **Bruno Pescara,** Rostock (Germany) **Cabinet of curiosities: Detecting and interpreting extraordinary variants** **Yvonne Kasmann,** Rostock (Germany)

Thursday, April 10th, 2025 – Young Investigator Day

	PART I & II – PRESENTATIONS OF THE FINALISTS: YOUNG INVESTIGATOR AWARD 2025 **Chairpersons: Anita Rauch,** Zurich **& Sven Cichon,** Basel
14:05 – 14:15	**Welcome address** **Anita Rauch,** Zurich **& Sven Cichon,** Basel
14:15 – 15:15	**SSMG Young Investigator Presentations with Q&A** (15 min each)
15:15 – 15:45	*Networking Break & Exhibition Visit*
15:45 – 16:45	**SSMG Young Investigator Presentations with Q&A** (15 min each)
16:45 – 17:45	*Poster Walk, Exhibition & Cocktail Reception*
	PART III – INVITED EVENING LECTURE **Chairpersons: Anita Rauch,** Zurich
17:45 – 18:30	**Beyond the genome: human genetics in the post-genomic era** **Malte Spielmann,** University Hospital Schleswig-Holstein
18:30 – 18:35	**Closing Remarks Day 1** **Sven Cichon,** Basel
From 20:00	*Dinner at the Sorell Hotel Zürichberg*

Friday, April 11th, 2025 – Scientific Meeting Day

	Chairperson: Anita Rauch, Zurich
09:00 – 09:15	**Opening and Welcome address** **Anita Rauch,** Zurich **& Ruxandra Bachmann-Gagescu,** Zurich
09:15 – 09:25	**Announcement of the Winner of the SSMG Young Investigator Award 2025** **Sven Cichon,** Basel
09:25 – 10:10	**New mutations, selfish testes and human disease** **Anne Goriely,** University of Oxford
10:10 – 10:55	**From rare to common: How the genetic architecture of kidney disease matters** **Olivier Devuyst,** University of Zurich
10:55 – 11:15	*Break*
11:15 – 12:00	**Into the unknown: long reads at the end of the odyssey** **Paranchai Boonsawat,** University of Zurich
12:00 – 12:30	**Satellite Symposium organized by AstraZeneca AG** **Expanding the BRCAness Horizon: AI-Aided Patient Identification and PARPi Applications in Emerging Cancer Types** **Lara Planas Paz,** Zurich
12:30 – 13:00	**Satellite Symposium organized by Oxford Nanopore Technologies plc** **Targeted nanopore sequencing approaches for analysing repeat expansion disorders, muscular dystrophies, and structural variants of unknown significance** **Morghan Lucas,** Munich
13:00 – 14:00	*Lunch Break and end of the Annual Meeting 2025*

Friday, April 11th, 2025 – Swiss Dysmorphology Meeting

	Chairpersons: Claudine Rieubland, Sion **& Julie De Geyter,** Basel
14:00 – 17:00	**Swiss Dysmorphology Meeting**

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Thursday, April 10th, 2025 – Oral presentation

Approved

14:15 – 14:30 h	Oral Presentation 1 Establishing Zebrafish and iPSCs-models of ANO4 missense variants linked to neurodevelopmental and epileptic disorders **Sissy Bassani,** University of Zurich
14:30 – 14:45 h	Oral Presentation 2 Moyamoya angiopathy in pediatric European patients: novel candidate genes and their associated biological roles **Elena Cabello,** University of Zurich
14:45 – 15:00 h	Oral Presentation 3 De novo variants in LDB1 cause a neurodevelopmental disorder **Rebecca Fluri,** University of Bern
15:00 – 15:15 h	Oral Presentation 4 Identification of novel candidate genes for inherited retinal disorders in 609 consanguineous families from Iran and Pakistan **Helen Frederiksen,** University of Lausanne
15:15 – 15:45 h	*Short Break*
15:45 – 16:00h	Oral Presentation 5 Systematic search for modifier genes influencing disease severity in families with hereditary angioedema **Asensio Gonzalez,** University Hospital Basel and University of Basel Department of Biomedicine
16:00 – 16:15 h	Oral Presentation 6 Biallelic missense variants in CSMD2 are associated with a neurodevelopmental disease and epilepsy **Clara Pailler,** University of Lausanne
16:15 – 16:30 h	Oral Presentation 7 De novo variants in LRRC8C resulting in constitutive channel activation cause a human multisystem disorder **Mathieu Quinodoz,** Institute of Molecular and Clinical Ophthalmology Basel
16:30 – 16:45 h	Oral Presentation 8 Bridging in silico predictions and functional validation: A zebrafish model for INPP5E variant classification for ciliopathy patients **Sydney Vibert,** University of Zurich

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Thursday, April 10th, 2025 – Poster presentation

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POSTER 1	Poster presentation 1 **SysNDD: An expert-curated database providing insights into the evolving landscape of neurodevelopmental disorders** **Simon Früh,** University of Bern
POSTER 2	Poster presentation 2 **UPDgraph: a bioinformatic tool to detect uniparental disomies (UPDs) using trio exome or genome sequencing data.** **Frederic Masclaux,** University Hospital of Geneva
POSTER 3	Poster presentation 3 **"Novel Insights into the Biomechanical Properties of the Aorta in Mice Modeling Hereditary Aortic Diseases"** **Janine Meienberg,** Center for Cardiovascular Genetics and Gene Diagnostics, Schlieren
POSTER 4	Poster presentation 4 **Heterozygous GAA case mimicking late-onset Pompe disease: In vitro assessment of the combination of genetic variants** **Magdalena Mroczek,** Center for Cardiovascular Genetics and Gene Diagnostics, Schlieren
POSTER 5	Poster presentation 5 **Identification of HIV restriction factors through mRNA expression variation in HIV-infected individuals** **Sergey Oreshkov,** CHUV Lausanne
POSTER 6	Poster presentation 6 **Biallelic protein coding variants in FTO cause a variable developmental phenotype: A genotype-phenotype correlation** **Radhakrishnan Periyasamy,** University Hospital Basel and University of Basel Department of Biomedicine
POSTER 7	Poster presentation 7 **Expanding the phenotypic spectrum of PAN2-related neurodevelopmental disorder: two siblings with a novel homozygous frameshift variant** **Violeta Rusu,** University Hospital of Bern
POSTER 8	Poster presentation 8 **Evaluation and Application of Long-Read Whole Genome Sequencing for SV Detection** **Giancarlo Tomio,** Center for Cardiovascular Genetics and Gene Diagnostics, Schlieren
POSTER 9	Poster presentation 9 **Cell-cell connectivity is impaired in keratinocytes with frameshift variants in keratin 10** **Agnieszka Tupalska,** University Hospital Basel and University of Basel Department of Biomedicine
POSTER 10	Poster presentation 10 **Functional characterization of BRCA1 and BRCA2 variants of unknown significance through semi-automated analysis of replication fork stability** **Stephanie van Gijn,** University of Zurich
POSTER 11	Poster presentation 11 **Prenatal diagnosis of MSL2-related ventriculomegaly in association with an inherited 15q13 microduplication** **Omar Zgheib,** University Hospital of Geneva

Sponsors

SATELLITES

AstraZeneca – Oxford Nanopore Technologies – varvis®

EXHIBITORS

Bencard GmbH – BGI Genomics Co Ltd – Chemie Brunschwig AG – Illumina Switzerland GmbH – Life & Brain – Macrogen Europe – Oxford Nanopore Technologies – PacBio – Roche Diagnostics (Schweiz) AG – SeQone S.A.S. – Sysmex – Thermo Fisher Scientific – Twist Bioscience – varvis® – Zytomed Systems GmbH – a ZYTOMICS company