



Swiss Society  
of Medical  
Genetics

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main program

# SSMG Annual Meeting 2025

WITH YOUNG INVESTIGATOR DAY

April 10<sup>th</sup> and 11<sup>th</sup>, 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

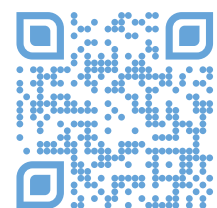
#### FAMH

Scientific programme SSMG meeting: 8 credits

### REGISTRATION

#### Medworld AG

Sennweidstrasse 46  
6312 Steinhausen



**Organizers: SSMG**

Björn Kleijkers | [bjoern.kleijkers@medgen.uzh.ch](mailto:bjoern.kleijkers@medgen.uzh.ch)

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



## Thursday, April 10<sup>th</sup>, 2025 – SSMG Business Meeting & General Assembly

Chairpersons: Isabel Filges, Basel & Thierry Nospikel, Geneva

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



## Thursday, April 10<sup>th</sup>, 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	<p><b>Welcome address</b>            Anita Rauch, Zurich &amp; Sven Cichon, Basel</p>
14:15 – 15:15	<b>SSMG Young Investigator Presentations with Q&amp;A (15 min each)</b>
15:15 – 15:45	<i>Networking Break &amp; Exhibition Visit</i>
15:45 – 16:45	<b>SSMG Young Investigator Presentations with Q&amp;A (15 min each)</b>
16:45 – 17:45	<i>Poster Walk, Exhibition &amp; Cocktail Reception</i>
	<b>PART III – INVITED EVENING LECTURE</b>
	Chairpersons: Anita Rauch, Zurich
17:45 – 18:30	<p><b>Beyond the genome: human genetics in the post-genomic era</b>            Malte Spielmann, University Hospital Schleswig-Holstein</p>
18:30 – 18:35	<p><b>Closing Remarks Day 1</b>            Sven Cichon, Basel</p>
From 20:00	<i>Dinner at the Sorell Hotel Zürichberg</i>

## Friday, April 11<sup>th</sup>, 2025 – Scientific Meeting Day

Chairperson: Anita Rauch, Zurich

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



## Friday, April 11<sup>th</sup>, 2025 – Swiss Dysmorphology Meeting

Chairpersons: Claudine Rieubland, Sion & Julie De Geyter, Basel

14:00 – 17:00	<b>Swiss Dysmorphology Meeting</b>
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**Thursday, April 10<sup>th</sup>, 2025 – Oral presentation**

 Not finally approved

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



**Thursday, April 10<sup>th</sup>, 2025 – Poster presentation**

▶ Not finally approved

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

## Sponsors

### SATELLITES

AstraZeneca – Oxford Nanopore Technologies – varvis®

### EXHIBITORS

Bencard GmbH – Chemie Brunschwig AG – Life & Brain – Macrogen Europe – PacBio – Roche Diagnostics (Schweiz) AG – SeQone S.A.S. – Sysmex – Thermo Fisher Scientific – Twist Bioscience – varvis® – Zytomed Systems GmbH – a ZYTOMICS company