Swiss Society of Medical Genetics

On site meeting

SSMG Annual Meeting 2024

WITH YOUNG INVESTIGATOR DAY

May 16th and 17th, 2024

Bern University Hospital, Auditorium Ettore Rossi

Conference Committee

Christiane Zweier
University Hospital of Bern

André Schaller
University Hospital of Bern

Keynote Speakers 2024

Thomas Bourgeron
Université Paris Cité:
Phenotypic effects of genetic variants associated with autism

Christa Flück
University Hospital of Bern:
Differences of Sex Development (DSD) – a clinical and genetic overview

Nadine Hornig
University of Kiel:
Molecular insights into androgen insensitivity

Anne Gregor
University Hospital of Bern:
Disease modeling in neurodevelopmental disorders

ORGANIZERS

Christiane Zweier
University Hospital of Bern

André Schaller
University Hospital of Bern

CREDIT POINTS

SGMG
8 CME points
Thursday/Friday

FAMH
8 CME points
Thursday/Friday

REGISTRATION

Medworld AG
Sennweidstrasse 46
6312 Steinhausen

https://sgmg.ch/de/meeting-2024

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PROGRAM

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Thursday, May 16th, 2024 – SSMG Business Meeting & General Assembly

Chairpersons: Isabel Filges, Basel & Naomi A. Porret, Bern

From 08:30	**Registration open**
09:45 – 12:15	**Business Meeting of the Swiss Society of Medical Genetics**
12:15 – 13:30	*Lunch, Poster and Exhibition Viewing*
13:30 – 14:00	**Lunch Symposium organized by AstraZeneca** **BRCAness and PARPi therapies** **Wiebke Solass,** University of Bern

Thursday, May 16th, 2024 – Young Investigator Day

	PART I & II – PRESENTATIONS OF THE FINALISTS: YOUNG INVESTIGATOR AWARD 2024 **Chairpersons: Christiane Zweier,** Bern **& Sven Cichon,** Basel
14:05 – 14:15	**Welcome address** **Christiane Zweier,** Bern **& Sven Cichon,** Basel
14:15 – 15:55	**SSMG Young Investigator Presentations with Q&A** (20 min each)
15:55 – 16:25	*Short Break*
16:25 – 17:45	**SSMG Young Investigator Presentations with Q&A** (20 min each)
	PART III – EVENING LECTURE **Chairpersons: André Schaller,** Bern
17:45 – 18:30	**Phenotypic effects of genetic variants associated with autism** **Thomas Bourgeron,** Paris
18:30 – 18:35	**Closing Remarks Day 1** **André Schaller,** Bern
From 18:35	*Apéro at the University Hospital of Bern*
From 20:00	*Dinner at Kornhauskeller in Bern*

Friday, May 17th, 2024- Scientific Meeting Day

	Chairperson: Christiane Zweier, Bern
08:20 – 08:50	**Satellite Symposium organized by Oxford Nanopore Technologies** **Rare Disease Diagnostics in Complex Genomic Regions with Long Reads** **Stephan Ossowski,** University Hospital Tübingen
09:00 – 09:15	**Opening and Welcome address** **Christiane Zweier,** Bern **& André Schaller,** Bern
09:15 – 09:25	**Announcement of the Winner of the SSMG Young Investigator Awards 2024** **Sven Cichon,** Basel
09:25 – 10:10	**Differences of Sex Development (DSD) – a clinical and genetic overview** **Christa Flück,** Bern
10:10 – 10:55	**Molecular insights into androgen insensitivity** **Nadine Hornig,** Kiel
10:55 – 11:15	*Break*
11:15 – 12:00	**Disease modeling in neurodevelopmental disorders** **Anne Gregor,** Bern
12:00 – 12:30	**Satellite Symposium organized by PacBio** **Resolving complex chromosomal rearrangement with PacBio Revio** **Lars Feuk,** Uppsala University
12:30 – 12:40	**Closing Remarks Day 2** **Christiane Zweier,** Bern **& André Schaller,** Bern
From 12:40	*Farewell Lunch*

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Thursday, May 16th, 2024 – Oral presentation

On site meeting

14:15 – 14:35 h	Oral Presentation 1 **Rare copy-number variants as modulators of common disease susceptibility** **Chiara Auwerx,** University of Lausanne
14:35 – 14:55 h	Oral Presentation 2 **Missense variants in ANO4 cause sporadic encephalopathic or familial epilepsy with evidence for a dominant-negative effect** **Anais Begemann,** University of Zurich
14:55 – 15:15 h	Oral Presentation 3 **Deleterious ZNRF3 germline variants as a novel cause of neurodevelopmental disorders with mirror brain phenotypes due to distinct domain-specific effects on Wnt/β-catenin signalinge** **Paranchai Boonsawat,** University of Zurich
15:15 – 15:35 h	Oral Presentation 4 **Deregulated of ion channels contribute to RHOBTB2 associated developmental and epileptic encephalopathy** **Franziska Langhammer,** University of Bern
15:35 – 15:55 h	Oral Presentation 5 **Bi-allelic variants in BRF2 are associated with perinatal death and craniofacial anomalies** **Francesca Mattioli,** University of Lausanne
15:55 – 16:25 h	*Short Break*
16:25 – 16:45 h	Oral Presentation 6 **Novel insights into tumorigenesis revealed by molecular analysis of Lynch syndrome cases with multiple colorectal tumors** **Alisa Olkinuora,** University Hospital Basel
16:45 – 17:05 h	Oral Presentation 7 **Biallelic missense variants in CSMD2 are associated with a neurodevelopmental disease and epilepsy** **Clara Pailler-Pradeau,** University of Lausanne
17:05 – 17:25 h	Oral Presentation 8 **Further characterization of BRSK2-associated neurodevelopmental disorder** **Palak Singhal,** University of Bern
17:25 – 17:45	Oral Presentation 9 **Long-read whole-genome sequencing analysis or rare structural risk variants in families with bipolar disorder** **Priyadarshini Thirunavukkarasu,** University Hospital Basel

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Thursday, May 16th, 2024 – Poster presentation

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POSTER 1	Poster presentation 1 **Structural and functional insights from single cell transcriptional profiles of pituitary tumors** **Maxime Brunner,** CHUV Lausanne
POSTER 2	Poster presentation 2 **Identification of microbiota components correlated with host lifestyle, molecular, biochemical, immunophenotypic measurements and genotype in a deeply phenotyped Sardinian cohort** **Maria Antonietta Diana,** CHUV Lausanne
POSTER 3	Poster presentation 3 **Current Genetic Counselling Practice in Paediatric Oncology in Switzerland A qualitative Study** **Corinne Gemperle,** University Children's Hospital Zurich
POSTER 4	Poster presentation 4 **Search for modifier genes influencing disease severity in Hereditary Angioedema** **Asensio Gonzalez,** University Hospital of Basel
POSTER 5	Poster presentation 5 **Health care transition to adulthood in young patients with rare genetic diseases: exploring the contributions of genetic counselling and genetic counsellors** **Sabrina Marti,** Cantonal hospital Aarau
POSTER 6	Poster presentation 6 **Long-read sequencing in SV calling and variant phasing** **Janine Meienberg,** Centre for Cardiovascular Genetics, Schlieren
POSTER 7	Poster presentation 7 **Genetic host factors in variation on viral control in HIV-infected individuals** **Sergey Oreshkov,** CHUV Lausanne
POSTER 8	Poster presentation 8 **Reclassification of variants of uncertain clinical significance in mendelian cardiomyopathies and arrythmias using targeted blood RNA analysis** **Thomas Rio Frio,** University Hospital of Geneva
POSTER 9	Poster presentation 9 **Further delineation of SCAF4 associated neurodevelopmental disorder** **Cosima Schmid,** University of Bern

Sponsors

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