



Swiss Society
of Medical
Genetics

Main programme



Register now

SSMG Annual Meeting 2026

WITH YOUNG INVESTIGATOR DAY

23rd and 24th April 2026

Centre Hospitalier Universitaire Vaudois (CHUV)
Auditoire César Roux

Conference Committee



**Cédric
Le Caignec**

Service de Médecine
Génétique, CHUV,
Lausanne



**Alexandre
Reymond**

Center for Integrative
Genomics, UNIL,
Lausanne



**Muhammad
Ansar**

Jules-Gonin Eye
Hospital, Fondation
Asile des Aveugles,
Lausanne

Keynote Speakers 2026



Lisenka Vissers

Radboud University, The Netherlands
Diagnosing with long reads



Christel Tran

University Hospital Lausanne
Metabolic disorders in adults



Zoltan Kutalik

University of Lausanne
Genetic architecture of complex
human traits



Christel Depienne

University Hospital Essen, Germany
Non-coding variants and diseases

CREDIT POINTS

SGMG

Business meeting of the SSMG: 2 credits
Scientific programme SSMG meeting: 9 credits
Swiss Dysmorphology meeting: 2 credits

FAMH

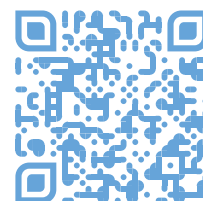
Scientific programme SSMG meeting: 7 credits

REGISTRATION

Medworld AG

Sennweidstrasse 46
6312 Steinhausen

registration@medworld.ch





Thursday, 23rd April, 2026 – SSMG Business Meeting / General Assembly

Chairpersons: Isabel Filges, Basel & Thierry Nospikel, Geneva

From 08:30 h	Registration open
10:00 – 12:00 h	Business Meeting of the Swiss Society of Medical Genetics
12:00 – 13:30 h	<i>Lunch, Poster and Exhibition Viewing</i>
13:30 – 14:00 h	Satellite Symposium Variant Interpretation: What Laboratories Are Actually Paying For Andreas Massouras, CEO Saphetor

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Thursday, 23rd April, 2026 – Young Investigator Day

14:00 – 14:05 h	Welcome address Cédric Le Caignec, Lausanne & Alexandre Reymond, Lausanne
	PART I – INVITED AFTERNOON LECTURE Chairperson: Cédric Le Caignec, Lausanne
14:05 – 14:50 h	Diagnosing with long reads Lisenka Vissers, Radboud University, Nijmegen, The Netherlands
	PART II – PRESENTATIONS OF THE YOUNG INVESTIGATOR AWARD 2026 FINALISTS Chairpersons: Cédric Le Caignec, Lausanne & Alexandre Reymond, Lausanne
14:50 – 15:50 h	4x SSMG Young Investigator Presentations with Q&A (15 min each)
15:50 – 16:10 h	<i>Networking Break & Exhibition Visit</i>
16:10 – 17:25 h	5x SSMG Young Investigator Presentations with Q&A (15 min each)
17:25 – 17:30 h	Wrap-up Day 1 Muhammad Ansar, Lausanne
17:30 – 18:30 h	<i>Poster walk and Apéro in Exhibition</i>
From 20:00 h	<i>Dinner</i>

Friday, 24th April, 2026 - Scientific Meeting Day

	PART III – SCIENTIFIC KEY LECTURES Chairpersons: Cédric Le Caignec, Lausanne & Alexandre Reymond, Lausanne
08:55 – 09:00 h	Opening and Welcome address Cédric Le Caignec, Lausanne & Alexandre Reymond, Lausanne
09:00 – 09:45 h	Adult inherited metabolic diseases: bridging metabolism and medical genetics Christel Tran, CHUV, Lausanne
09:45 – 09:55 h	Announcement of the Winner of the SSMG Young Investigator Award 2026 Sven Cichon, Basel
09:55 – 10:40 h	Genetic architecture of complex human traits Zoltan Kutalik, University of Lausanne, and University Center for Primary Care and Public Health, Lausanne
10:40 – 11:10 h	<i>Break</i>
11:10 – 11:55 h	Non-coding variants and diseases Christel Depienne, University Hospital Essen, Essen, Germany
12:00 – 12:30 h	Satellite Symposium Long-Read Sequencing in Clinical Genomics: New Opportunities for Precision Medicine Ole Halfdan Larsen, PhD, Aarhus Universitetshospital, Denmark
12:30 – 13:00 h	Satellite Symposia TruPath Genome in Rare Disease: Colocation-based Reconstruction of Complex Chromosomal Architecture Klaus Wagner, Medical University of Graz, Austria
13:00 – 14:00 h	<i>Lunch Break and end of the Annual Meeting 2026</i>

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Friday, 24th April, 2026 - Swiss Dysmorphology Meeting

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

14:00 – 16:00 h	Swiss Dysmorphology Meeting
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Thursday, 23rd April, 2026 – Oral presentation

14:50 – 15:05 h	<p>Oral Presentation 1 Compounding of rare copy-number variants and polygenic risk: A genetic signature of assortative mating Caterina Cevallos, University of Lausanne</p>
15:05 – 15:20 h	<p>Oral Presentation 2 Hypersynchronous neural network activity in a hiPSC-derived model of PSMD12-associated neurodevelopmental disorder is rescued by proteasome activation Simon Früh, University Hospital of Berne</p>
15:20 – 15:35 h	<p>Oral Presentation 3 MAZ, genome architecture and language disorders Aymeric Masson, University of Lausanne</p>
15:35 – 15:50 h	<p>Oral Presentation 4 Loss of function variants in SAXO6, encoding a microtubule inner protein of photoreceptor cilia, are a novel cause of late-onset retinal dystrophy Abigail Moyo, Institute of Molecular and Clinical Ophthalmology Basel</p>
15:50 – 16:10 h	<i>Short Break</i>
16:10 – 16:25 h	<p>Oral Presentation 5 Transcriptomic Dynamics During Minipuberty Reveal Divergent Developmental Programs in Preterm and Full-Term Infants Sergey Oreshkov, University of Lausanne</p>
16:25 – 16:40 h	<p>Oral Presentation 6 Biallelic missense variants in CSMD2 and its haploinsufficiency are associated with neurodevelopmental disease. Clara Pailler-Pradeau, University of Lausanne</p>
16:40 – 16:55 h	<p>Oral Presentation 7 Genetic Analysis of Visual Impairment in 638 Consanguineous Multiplex Families from Pakistan and Iran Abdur Rashid, University of Lausanne</p>
16:55 – 17:10 h	<p>Oral Presentation 8 Identifying and Characterizing Genetic Modifiers in Long QT Syndrome Joana Rechsteiner, University Hospital of Berne</p>
17:10 – 17:25 h	<p>Oral Presentation 9 Moving in uncharted territory: searching for disease-associated rare structural variants in multiplex families with bipolar disorder using long-read sequencing Priyadarshini Thirunavukkarasu, University of Basel</p>

Sponsors

SATELLITES

AstraZeneca – Oxford Nanopore Technologies – varvis®

EXHIBITORS

Thermo Fisher Scientific – Ultragenyx – Agilent – Illumina – Devyser – Qiagen AG – PacBio – varvis – Sophia Genetics – Bencard AG – Sysmex Suisse AG – Roche Diagnostics (Schweiz) AG – LIFE & BRAIN GmbH – Twist Bioscience – SeqOne – Element Biosciences – Integra – Oxford Nanopore Technologies


Thursday, 23rd April, 2026 – Poster presentation
 not yet confirmed

POSTER 1	The localisation of variant impacts the phenotype of patients with KRT10-nEDD-revertant-mosaic Bettina Burger, University of Basel
POSTER 2 withdrawn	Withdrawn by author
POSTER 3	First Prenatal Presentation of a Biallelic truncating PLAA Variant: Expanding the Phenotypic Spectrum of PLAA-Related Neurodevelopmental Disorders Yasmine El Ayeb, Centre Hospitalier Universitaire Vaudois
POSTER 4	Molecular characterisation of a novel homozygous PPOX splicing variant causing Variegate Porphyria with brachydactyly and osteolysis in a single family Rosalinda Giannini, Hôpitaux Universitaires de Genève
POSTER 5	Inherited cancer syndromes in adult oncology: a referral guide based on malignant tumor and polyp features Audrey Guilmot, Centre Hospitalier Universitaire Vaudois
POSTER 6	Utilizing Long Read Sequencing for Comprehensive Detection of Copy Number Aberrations in Hematological Malignancies Trung Hieu Luu, Centre Hospitalier Universitaire Vaudois
POSTER 7	Comparative analysis of genotyping and low-pass sequencing for the identification of pharmacogenetic variants Flavia Hodel, Centre Hospitalier Universitaire Vaudois
POSTER 8	When normal biochemistry is misleading: a novel CYP27B1 variant identified in a family with longstanding unexplained rickets Summer Hofmann, University Hospital of Berne
POSTER 9	Biallelic MYH3 variants cause distal arthrogyrosis in compound heterozygosity and a subclinical phenotype in simple heterozygosity. Codominance or recessive inheritance? Philippe Khau Van Kein, Hôpitaux Universitaires de Genève
POSTER 10	Variable Hypertrophic Cardiomyopathy Severity in Siblings Carrying a Homozygous MYBPC3 Variant. Philippe Khau Van Kein, Hôpitaux Universitaires de Genève
POSTER 11	Phenotypic cluster analysis of genetically undiagnosed cases of differences of sex development Chrysanthi Kouri, University Hospital of Berne
POSTER 12	Germline TUBB Variant Associated with Bone Marrow Failure and Somatic 6p Loss of Heterozygosity: Expanding the Phenotypic Spectrum of Tubulinopathies Henri Margot, Hôpitaux Universitaires de Genève
POSTER 13	Preclinical Evidence for Manufacturer-Specific Effects of Celiprolol in a vEDS Mouse Model Janine Meienberg, Centre for Cardiovascular Genetics and Genetic Diagnostics, Schlieren
POSTER 14	Autonomous at 71: Late Diagnosis of Kabuki Syndrome Iris Peters, Hôpitaux Universitaires de Genève
POSTER 15	Genotype-phenotype correlation and molecular dynamics simulation for FTO variants causing a variable neurodevelopmental disorder Periyasamy Radhakrishnan, University of Basel
POSTER 16	Perspectives of Health Care Professionals Involved in Prenatal Genetic Testing in Switzerland Kirsten Riggan, ETH Zurich
POSTER 17	Does using Perplexity AI change the performance of genetic counselling case preparation? Lisa Schulze, University Hospital Basel
POSTER 18	Allelic Imbalance in PacBio HiFi Sequencing Challenges Clinical Variant Detection Despite Read Length Giancarlo Tomio, Centre for Cardiovascular Genetics and Genetic Diagnostics, Schlieren
POSTER 19	Biallelic loss-of-function and missense variants in novel genes associated with neurodevelopmental disorders Mukhtar Ullah, Institute Of Molecular And Clinical Ophthalmology Basel
POSTER 20	GenMasterAI: A Local Large Language Model-Based Agentic and Reasoning software to Democratize Genetic Analysis Jing Zhai, Lausanne University Hospital