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

www.sgmg.ch

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

Chairpersons: Isabel Filges, Basel & Thierry Nouspikel, 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	*Lunch, Poster and Exhibition Viewing*
13:30 – 14:00 h	**Satellite Symposia** organised by SAPHETOR **Variant Interpretation: What Laboratories Are Actually Paying For** **Andreas Massouras,** CEO Saphetor

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	*Networking Break & Exhibition Visit*
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	*Poster walk and Apéro in Exhibition*
From 20:00 h	*Dinner*

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 **& Muhammad Ansar,** 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	*Break*
11:10 – 11:55 h	**Non-coding variants and diseases** **Christel Depienne,** University Hospital Essen, Essen, Germany
12:00 – 12:30 h	**Satellite Symposia** organised by Oxford NANOPORE Technologies **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** organised by illumina **TruPath Genome in Rare Disease: Colocation-based Reconstruction of Complex Chromosomal Architecture** **Klaus Wagner,** Medical University of Graz, Austria
13:00 – 14:00 h	*Lunch Break and end of the Annual Meeting 2026*

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	Oral Presentation 1 **Compounding of rare copy-number variants and polygenic risk: A genetic signature of assortative mating** **Caterina Cevallos,** University of Lausanne
15:05 – 15:20 h	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
15:20 – 15:35 h	Oral Presentation 3 **MAZ, genome architecture and language disorders** **Aymeric Masson,** University of Lausanne
15:35 – 15:50 h	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 Moye,** Institute of Molecular and Clinical Ophthalmology Basel
15:50 – 16:10 h	*Short Break*
16:10 – 16:25 h	Oral Presentation 5 **Transcriptomic Dynamics During Minipuberty Reveal Divergent Developmental Programs in Preterm and Full-Term Infants** **Sergey Oreshkov,** University of Lausanne
16:25 – 16:40 h	Oral Presentation 6 **Biallelic missense variants in CSMD2 and its haploinsufficiency are associated with neurodevelopmental disease.** **Clara Pailler-Pradeau,** University of Lausanne
16:40 – 16:55 h	Oral Presentation 7 **Genetic Analysis of Visual Impairment in 638 Consanguineous Multiplex Families from Pakistan and Iran** **Abdur Rashid,** University of Lausanne
16:55 – 17:10 h	Oral Presentation 8 **Identifying and Characterizing Genetic Modifiers in Long QT Syndrome** **Joana Rechsteiner,** University Hospital of Berne
17:10 – 17:25 h	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

Sponsors

SATELLITES

Illumina – Oxford Nanopore Technologies – Saphetor®

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 Boiscience – SeqOne – Element Biosciences – Integra – Oxford Nanopore Technologies

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Thursday, 23rd April, 2026 – Poster presentation

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 arthrogryposis 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