Swiss Society of Medical Genetics

ONLINE VENUE

PROGRAM

SSMG Annual Meeting 2021

YOUNG INVESTIGATOR DAY

January 21st and 22nd, 2021

www.sgmg.ch/meeting-2021

ORGANIZERS

Anita Rauch
Institute of Medical Genetics, University of Zurich

Björn Kleijkers
Institute of Medical Genetics, University of Zurich

Sven Cichon
Institute of Medical Genetics and Pathology, University of Basel

REGISTRATION FEES

SSMG-Member
CHF 100.–
Non-Member
CHF 175.–
Trainee/PhD student
CHF 50.–
General Assembly:
For members only
Free of charge

CME CREDIT POINTS

FMH: 3 CME points/day

REGISTRATION

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SUPPORT

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January 21st, 2021 – Young Investigator Day — ONLINE VENUE

PART I – PRESENTATIONS OF THE FINALISTS FOR THE YOUNG INVESTIGATOR AWARD 2020

Chairpersons Sven Cichon, Basel, Anita Rauch Zurich

14.05 – 14.15 h	Welcome Address **Anita Rauch,** Zurich / **Sven Cichon,** Basel
14.15 – 16.00 h	Oral Presentations 1–7 with Q&A (15 min. each) for details see page 3
16.00 – 16.30 h	*Break*

PART II – PRESENTATIONS OF ABSTRACTS

16.30 – 18.00 h	Oral Presentations 8–13 with Q&A (15 min. each) for details see page 3
18.00 – 18.30 h	*Break*

January 21st, 2021 – Evening Lecture — ONLINE VENUE

PART III – INVITED EVENING LECTURE

Chairperson Anita Rauch, Zurich

18.30 – 19.30 h	Polygenic Risk Models – what are they and are they ready for the clinic? **Cecile J.W. Janssens,** Atlanta USA
19.30 – 19.35 h	Closing Remarks Day 1 **Anita Rauch,** Zurich

January 22nd, 2021 – Scientific & Business Meeting Day — ONLINE VENUE

09.00 – 09.15 h	Opening and Welcome Address **Anita Rauch,** Zurich / **Sven Cichon,** Basel
09.15 – 09.25 h	Announcement of the Winner of the SSMG Young Investigator Award 2020
09.25 – 10.10 h	Gene Therapy for Monogenetic Diseases of the Immune System **Janine Reichenbach,** Zurich
10.10 – 10.55 h	Ribosomal DNA – an epigenetic sensor of gene-environment interactions **Vardhman Rakyan,** London UK
10.55 – 11.25 h	*Short Break*
11.25 – 12.00 h	Current challenges in Mendelian Genetics: variant interpretation, oligogenicity, phenotypic variability and genetic modifiers – lessons learned from the ciliopathies **Ruxandra Bachmann-Gagescu,** Zurich
12.00 – 12.30 h	Satellite Symposia – organized by Novartis A new vision in retinal gene therapy: From genetic diagnosis to treatment **Hendrik Scholl,** Professor and Chairman Department of Ophthalmology, University of Basel, Director Institute of Molecular and Clinical Ophthalmology Basel (IOB) **Carlo Rivolta,** Head of the Ophthalmic Genetics Group at Institute of Molecular and Clinical Ophthalmology Basel (IOB), University of Basel
12.30 – 13.00 h	Satellite Symposia – organized by Roche Molecular Tumor Profiling and Incidental Germline Findings **Chantal Pauli,** University Hospital Zurich, Institut for Pathology and Molecular Biology
13.00 – 14.30 h	*Lunchtime*

January 22nd, 2021 – SSMG General Assembly: For members only — ONLINE VENUE

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

14.30 – 17.00 h	**Business Meeting of the Swiss Society of Medical Genetics**

January 21st, 2021 – ORAL PRESENTATIONS

ONLINE VENUE

14.15 – 14.30 h	Oral Presentation 1 **Variants in the USP48 ubiquitin hydrolase are associated with Autosomal Dominant Non-Syndromic Hereditary Hearing Loss** **Sissy Bassani**, Lausanne
14.30 – 14.45 h	Oral Presentation 2 **New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics** **Anais Begemann**, Zurich
14.45 – 15.00 h	Oral Presentation 3 **Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders: a apparently non-genetic phenocopy revealing a possible role for cartilage stem cells** **Lorenzo Botto**, Utah USA
15.00 – 15.15 h	Oral Presentation 4 **Tanycytes, ependymal cells and Co. A single cell analysis of brain third ventricle** **Maxime Brunner**, Lausanne
15.10 – 15.30 h	Oral Presentation 5 **Whole genome sequencing: why does it help?** **Katarina Cisarova**, Lausanne
15.30 – 15.45 h	Oral Presentation 6 **Medical Therapy of Vascular Ehlers-Danlos Syndrome: Challenging the Paradigm of Interchangeable Antihypertensive Drugs** **Nicolo Dubacher**, Zurich
15.45 – 16.00 h	Oral Presentation 7 **Newborn screening for Severe Combined Immunodeficiency (SCID) using combined cell receptor excision circles (TREC)/kappa-deleting element recombination element (KREC) assays and next generation sequencing: diagnostic yield from the newly established Swiss programme** **Magdeldin Elgizouli**, Zurich
16.00 – 16.30 h	*Break*
16.30 – 16.45 h	Oral Presentation 8 **Atonal homolog 7 as molecular basis for optic nerve hypoplasia and other retinal diseases** **David Grubich Atac**, Zurich
16.45 – 17.00 h	Oral Presentation 9 **SwissGenVar: A genetic data-sharing platform and knowledge-database for harmonization and up-scaling of clinical grade interpretation of genetic variants to foster personalized health care in Switzerland** **Dennis Kraemer**, Zurich
17.00 – 17.15 h	Oral Presentation 10 **Long-range PCR-based NGS applications to diagnose Mendelian retinal diseases** **Jordi Maggi**, Zurich
17.15 – 17.30 h	Oral Presentation 11 **Investigating the role of the Bardet-Biedl protein Bbs1 in zebrafish using –omics approaches** **Markus Masek**, Zurich
17.30 – 17.45 h	Oral Presentation 12 **Differentially expressed genes reflect disease-induced rather than disease-causing changes in the transcriptome** **Eleonora Porcu**, Lausanne
17.45 – 18.00 h	Oral Presentation 13 **CoverageMaster: a clinical grade and user oriented CNV caller** **Melivoia Rapti**, Lausanne

January 21st – 22nd, 2021 – POSTERS PRESENTATIONS

ONLINE VENUE

POSTER 2	**Intellectual disability, immune deficiency, autoimmunity, cutis marmorata and stroke: a pleiotropic disorder in four individuals associated with a Swiss founder mutation in the TPP2 gene** **Maria Isis Atallah**, Lausanne
POSTER 3	**Exome sequencing in prenatal diagnosis: results from 74 cases** **Xavier Blanc**, Geneva
POSTER 4	**Genetic findings in a Swiss cohort of patients with palmoplantar keratoderma** **Bettina Burger**, Basel
POSTER 5	**Biallelic loss of function variants in RFLNA and spondylo-carpal-tarsal synostosis syndrome** **Belinda Campos-Xavier**, Lausanne
POSTER 6	**New Insights into Clinical Whole-Genome Sequencing: Co-Occurring Rare Diseases and Pharmacogenetic Profiling** **Sylvan Caspar**, Zurich
POSTER 7	**PGT-M for a de novo case of Hereditary Multiple Exostoses (HME)** **Nadia Fiandanese**, Bioggio
POSTER 8	**Gene ontology enrichment analysis of renal agenesis: improving prenatal molecular diagnosis** **Silvia Kalantari**, Turin IT
POSTER 9	**Whole Genome Sequencing reveals bi-allelic ERGIC1 deletion in mild congenital arthrogryposis** **Caterina Marconi**, Geneva
POSTER 10	**Targeted prenatal exome sequencing in a series of fetal structural anomalies detected by ultrasonography: first experience of the University Hospitals of Geneva** **Lina Quteineh**, Geneva
POSTER 11	**Variants in calcium channel genes (CACNA1A, CACNA1C, CACNA1E) associated with noncanonical phenotypes: a common theme?** **Beryl Royer-Bertrand**, Lausanne
POSTER 12	**Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype** **Paolo Zanoni**, Zurich