Swiss Society of Medical Genetics

Online

SSMG Annual Meeting 2022

WITH YOUNG INVESTIGATOR DAY

April 7th and 8th, 2022

ORGANIZERS

Marc Abramowicz
University Hospital of Geneva HUG

Thierry Nouspikel
University Hospital of Geneva HUG

Sven Cichon
University of Basel

CREDIT POINTS

FMH: 9 CME points
Thursday/Friday

FAMH: 7 CME points
Thursday/Friday

REGISTRATION

Medworld AG
Sennweidstrasse 46
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https://sgmg.ch/meeting-2022

https://sgmg.ch/de/meeting-2022

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Thursday, April 7th, 2022 – Young Investigator Day

	PART I & II – PRESENTATIONS OF THE FINALISTS: YOUNG INVESTIGATOR AWARD 2022 **Chairpersons: Marc Abramowicz,** Geneva & **Sven Cichon,** Basel
14.05 – 14.15 h	Welcome Address **Marc Abramowicz,** Geneva
14.15 – 16.00 h	SSMG Young Junior Investigator Presentations with Q&A (15 min. each)
16.00 – 16.20 h	*Short Break*
16.20 – 17.35 h	SSMG Young Junior Investigator Presentations with Q&A (15 min. each)
17.35 – 18.10 h	*Short Break*
	PART III – INVITED EVENING LECTURE **Chairperson: Thierry Nouspikel,** Geneva & **Marc Abramowicz,** Geneva
18.10 – 18.55 h	Somatic mutation and clonal expansions in normal tissues **Iñigo Martincorena,** Cambridge
18.55 – 19.00 h	Closing Remarks Day 1 **Marc Abramowicz,** Geneva

Friday, April 8th, 2022 – Scientific & Business Meeting Day

	Chairperson: Marc Abramowicz, Geneva
09.00 – 09.15 h	Opening and Welcome Address **Marc Abramowicz,** Geneva
09.15 – 09.25 h	Announcement of the Winner of the SSMG Young Investigator Award 2022 **Sven Cichon,** Basel
09.25 – 10.10 h	Neurons from stem cells for Huntington's disease research **Elena Cattaneo,** Milan
10.10 – 10.55 h	Neurodevelopmental disorders: genotypes, phenotypes and function **Christiane Zweier,** Bern
10.55 – 11.15 h	*Break*
11.15 – 12.00 h	NIPD-M: non-invasive prenatal diagnostic for monogenic diseases **Thierry Nouspikel,** Geneva
12.00 – 13.30 h	*Lunch Break*

Friday, April 8th, 2022 – SSMG General Assembly
Chairpersons: Isabel Filges, Basel & Naomi A. Porret, Bern

13.30 – 16.00 h	**Business Meeting of the Swiss Society of Medical Genetics**

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PROGRAM

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Thursday, April 7th, 2022 – Oral Presentation

ONLINE VENUE

14.15 – 14.30 h	Oral Presentation 1 **The individual and global impact of copy number variants on complex traits** **Chiara Auwerx**, Lausanne
14.30 – 14.45 h	Oral Presentation 2 **Single cell Transcriptomics of Pituitary Neuroendocrine Tumors (PitNETs)** **Maxime Brunner**, Lausanne
14.45 – 15.00 h	Oral Presentation 3 **Comparison of neurotransmitter gene expression with related receptor densities in human hippocampal regions** **Bettina Burger**, Basel
15.00 – 15.15 h	Oral Presentation 4 **FBXO11 haploinsufficiency also stems from de novo missense variants and impairs neuronal differentiation and migration in an iPSC-based neuronal model** **Anne Gregor**, Bern
15.15 – 15.30 h	Oral Presentation 5 **Mutation landscape in 155 children with Developmental and Epileptic Encephalopathies recruited from three University hospital centers** **Eva Hammar**, Geneva
15.30 – 15.45 h	Oral Presentation 6 **Current Benefit of Whole Genome Sequencing Compared to Whole Exome Sequencing in Patients with Developmental and Epileptic Encephalopathies** **Ivan Ivanovski**, Zurich
15.45 – 16.00 h	Oral Presentation 7 **Genotype-phenotype correlations and pathomechanisms in RHOBTB2-associated developmental and epileptic encephalopathy** **Franziska Langhammer**, Bern
16.00 – 16.20 h	*Break*
16.20 – 16.35 h	Oral Presentation 8 **Identification of novel genetic causes of intellectual disability in consanguineous families** **Francesca Mattioli**, Lausanne
16.35 – 16.50 h	Oral Presentation 9 **How protein expression links somatic mutations to tumor phenotypes in chronic lymphocytic leukemia** **Fabienne Meier-Abt**, Zurich
16.50 – 17.05 h	Oral Presentation 10 **A polygenic risk score to predict sudden cardiac arrest in patients with cardiovascular disease** **Eleonora Porcu**, Lausanne
17.05 – 17.20 h	Oral Presentation 11 **Beyond clinical NGS: RNA-based analysis to overcome uncertainty** **Thomas Rio Frio**, Geneva
17.20 – 17.35 h	Oral Presentation 12 **Distribution analysis of missense variants in the human genome reveals widespread gene-specific clustering patterns and improves prediction of pathogenicity** **Mathieu Quinodoz**, Basel

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Thursday, April 7th, 2022 – Poster presentation — ONLINE VENUE

POSTER 1	Genetic testing for cerebellar ataxia, neuropathy and vestibular areflexia syndrome (CANVAS) by repeat expansion analyses in the RFC1 gene	**Urs Graf**, Zurich
POSTER 2	Segmental uniparental isodisomy in combination with a novel DOCK7 variant associated with infantile epileptic encephalopathy and cortical visual impairment	**Fatma Kivrak Pfiffner**, Zurich
POSTER 3	Phenotypic and molecular analysis of RP1 variants show intrafamilial variability and protein misslocalization	**Samuel Koller**, Zurich
POSTER 4	You cannot interpret what you do not detect: How not to miss clinically-relevant ClinVar and HGMD variants	**Janine Meienberg**, Schlieren
POSTER 5	The « Amish » c.3330+2T>G splice variant in MYBPC3 associated with hypertrophic cardiomyopathy is an ancient Swiss founder mutation.	**Claire Redin**, Lausanne
POSTER 6	Novel homozygous variant in SERPING1 causes hereditary angioedema in a consanguineous Brazilian family	**Luana Sella Motta Maia**, Basel
POSTER 7	The Challenging Choice of Gene Panel Size: Our Experience with Hypertrophic Cardiomyopathy.	**Sebastian Suchet**, Geneva
POSTER 8	Detection of KRT10 expression at low level in the human epidermal basal layer	**Agnieszka Tupalska**, Basel
POSTER 9	The genetic landscape and clinical implication of pediatric Moyamoya angiopathy in a multiethnic cohort	**Paolo Zanoni**, Zurich

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