



# SSMG Annual Meeting 2023

WITH YOUNG INVESTIGATOR DAY

April 27<sup>th</sup> and 28<sup>th</sup>, 2023

Altes Kraftwerk, Basel

## Conference committee



**Isabel Filges**

Medical Genetics, University  
Hospital Basel and University  
of Basel



**Sven Cichon**

Medical Genetics, University  
Hospital Basel and University  
of Basel

## Keynote Speakers 2023



**Richard Neher**

Biozentrum University of Basel:  
Evolutionary aspects of SARS-  
CoV2



**Diane van Opstal**

Erasmus MC, Netherlands:  
Experiences with genome-wide  
non-invasive prenatal testing



**Anita Rauch**

University of Zurich:  
Carrier screening



**Simon N. Waddington**

University College London:  
Gene therapy: clinical reality  
and experimental work

### ORGANIZERS

**Isabel Filges**

University of Basel

**Sven Cichon**

University of Basel

### CREDIT POINTS

**FAMH:**

8 CME points  
Thursday/Friday

**FMH:**

8 CME points  
Thursday/Friday

### REGISTRATION

**Medworld AG**

Sennweidstrasse 46  
6312 Steinhausen



<https://sgmg.ch/de/meeting-2023>



## Thursday, April 27<sup>th</sup>, 2023 – Young Investigator Day

**PART I & II – PRESENTATIONS OF THE FINALISTS: YOUNG INVESTIGATOR AWARD 2023**  
Chairpersons: Isabel Filges, Basel & Sven Cichon, Basel

14.05 – 14.15 h	<b>Welcome Address</b> Isabel Filges, Basel & Sven Cichon, Basel
14.15 – 15.55 h	<b>SSMG Young Junior Investigator Presentations with Q&amp;A</b> (15 min. each)
15.55 – 16.25 h	<i>Short Break</i>
16.25 – 17.25 h	<b>SSMG Young Investigator Presentations with Q&amp;A</b> (15 min. each)

**PART III – INVITED EVENING LECTURE**  
Chairperson: Sven Cichon, Basel

17.25 – 18.10 h	<b>From wave to wave: rapid adaptation and immune escape of SARS-CoV-2</b> Richard Neher, Basel
18.10 – 18.15 h	<b>Closing Remarks Day 1</b> Isabel Filges, Basel
<i>Evening</i>	<i>Apéro riche</i>

## Friday, April 28<sup>th</sup>, 2023 – Scientific & Business Meeting Day

Chairperson: Isabel Filges, Basel

09.00 – 09.15 h	<b>Opening and Welcome Address</b> Isabel Filges, Basel & Sven Cichon, Basel
09.15 – 09.25 h	<b>Announcement of the Winner of the SSMG Young Investigator Award 2023</b> Sven Cichon, Basel
09.25 – 10.10 h	<b>Experiences with genome-wide non-invasive prenatal testing</b> Diane van Opstal, Netherlands
10.10 – 10.55 h	<b>Gene therapy.. at last</b> Simon N. Waddington, London
10.55 – 11.15 h	<i>Break</i>
11.15 – 12.00 h	<b>Carrier screening</b> Anita Rauch, Zurich
12.00 – 12.30 h	Satellite Symposia organized by <b>AADC deficiency and molecular Background</b> Nenad Blau, University Children's Hospital Zurich <b>DDC Variants and Classification</b> Nastassja Himmelreich, University Hospital Heidelberg
12.30 – 13.00 h	Satellite Symposia organized by Astra Zeneca <b>Therapeutic implications of gBRCA1/2 testing in HER2-negative breast cancer</b> Cornelia Leo, Hospital of Baden
13.00 – 14.30 h	<i>Lunch Break</i>



## April 28<sup>th</sup>, 2023 – SSMG Business Meeting / General Assembly

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

14.30 – 17.00 h	<b>Business Meeting of the Swiss Society of Medical Genetics</b>
-----------------	--



Thursday, April 27<sup>th</sup>, 2023 – Oral presentation

On site meeting

14.15 – 14.35 h	Oral Presentation 1 <b>Mutation-specific pathophysiological mechanisms of AFF3</b> Sissy Bassani, Center of Integrative Genomics, Lausanne
14.35 – 14.55 h	Oral Presentation 2 <b>The role of ATOH7 in neuroretinal development modelled in human iPSC-derived retinal organoids</b> David Grubich Atac, University of Zurich
14.55 – 15.15 h	Oral Presentation 3 <b>Added Value of Statins in the Drug Therapy of Vascular Ehlers-Danlos Syndrome</b> Janine Meienberg, Centre for Cardiovascular Genetics, Schlieren
15.15 – 15.35 h	Oral Presentation 4 <b>Cracking the Code: Genetic Insights from the Swiss Kidney Stone Cohort</b> Johannes Münch, University of Zurich
15.35 – 15.55 h	Oral Presentation 5 <b>Clinical utility of periconception carrier screening by genome sequencing</b> Boonsawat Paranchai, University of Zurich
15.55 – 16.25 h	<i>Short Break</i>
16.25 – 16.45 h	Oral Presentation 6 <b>LHX2 haploinsufficiency causes a variable neurodevelopmental disorder</b> Cosima Schmid, University of Bern
16.45 – 17.05 h	Oral Presentation 7 <b>In-silico approach to identify novel susceptibility genes for bipolar disorder involved in neurodevelopment</b> Priyadarshini Thirunavukkarasu, University Hospital Basel
17:05 – 17.25 h	Oral Presentation 8 <b>Application of artificial intelligence aided diagnostics to the clinical differentiation between Constitutional delay of growth and puberty and congenital hypogonadotropic hypogonadism emphasizes the relevance of polygenic scores based on rare variants</b> Yassine Zouaghi, CHUV Lausanne



Thursday, April 27<sup>th</sup>, 2023 – Poster presentation

▶ On site meeting

POSTER 1	Poster presentation 1 <b>"Structural and functional insights from single cell transcriptional profiles of pituitary tumors"</b> Maxime Brunner, CHUV Lausanne
POSTER 2	Poster presentation 2 <b>"Long-read Nanopore-based sequencing to sequence the whole CFTR gene on Cystic Fibrosis African patients (Case of three Moroccan patients)"</b> Nada El Makhzen, University of Bern
POSTER 3	Poster presentation 4 <b>Two in one: WGS-based personalized PGx profiling</b> Janine Meienberg, Centre for Cardiovascular Genetics, Schlieren
POSTER 4	Poster presentation 5 <b>The interplay between gene expression in blood and spontaneous HIV control</b> Sergey Oreshkov, CHUV Lausanne
POSTER 5	Poster presentation 6 <b>OGM reveals the complexity of two constitutional structural chromosomal events</b> Lina Quteineh, University Hospital of Geneva
POSTER 6	Poster presentation 7 <b>Re-evaluation of variants of uncertain clinical significance in hypertrophic and dilated cardiomyopathy: the experience of the University Hospital of Geneva</b> Sebastian Suchet, University Hospital of Geneva

## Sponsors

### EXHIBITORS

Blueprint Genetics  
 ElementBiosciences  
 Life&Brain GmbH  
 PacBio  
 PTC Therapeutics  
 Sysmex Suisse AG  
 Vaudaux-Eppendorf AG

### SATELLITES

AstraZeneca  
 PTC Therapeutics

## Industry partner

