SSMG Annual Meeting 2021
YOUNG INVESTIGATOR DAY
January 21\textsuperscript{st} and 22\textsuperscript{nd}, 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**

www.sgmg.ch/meeting-2021

**SUPPORT**

Medworld AG  
Sennweidstrasse 46  
6312 Steinhausen  
registration@medworld.ch  
Helpline  
Tel +41 41 748 23 00
January 21st, 2021 – Young Investigator Day

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

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

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

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

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# ORAL PRESENTATIONS

**January 21st, 2021**

<table>
<thead>
<tr>
<th>Time</th>
<th>Presentation</th>
<th>Title</th>
<th>Speaker/Location</th>
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<tbody>
<tr>
<td>14.15</td>
<td>Oral 1</td>
<td>Variants in the USP48 ubiquitin hydrolase are associated with Autosomal Dominant Non-Syndromic Hereditary Hearing Loss</td>
<td>Sissy Bassani, Lausanne</td>
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<tr>
<td>14.30</td>
<td>Oral 2</td>
<td>New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics</td>
<td>Anais Begemann, Zurich</td>
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<tr>
<td>14.45</td>
<td>Oral 3</td>
<td>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</td>
<td>Lorenzo Botto, Utah USA</td>
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<tr>
<td>15.00</td>
<td>Oral 4</td>
<td>Tanyctyes, ependymal cells and Co. A single cell analysis of brain third ventricle</td>
<td>Maxime Brunner, Lausanne</td>
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<tr>
<td>15.10</td>
<td>Oral 5</td>
<td>Whole genome sequencing: why does it help?</td>
<td>Katarina Cisarova, Lausanne</td>
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<tr>
<td>15.30</td>
<td>Oral 6</td>
<td>Medical Therapy of Vascular Ehlers-Danlos Syndrome: Challenging the Paradigm of Interchangeable Antihypertensive Drugs</td>
<td>Nicolet Dubacher, Zurich</td>
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<td>15.45</td>
<td>Oral 7</td>
<td>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</td>
<td>Magdeldin Elgizouli, Zurich</td>
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<td>16.00</td>
<td>Break</td>
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<tr>
<td>16.30</td>
<td>Oral 8</td>
<td>Atonal homolog 7 as molecular basis for optic nerve hypoplasia and other retinal diseases</td>
<td>David Grubich Atac, Zurich</td>
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<tr>
<td>16.45</td>
<td>Oral 9</td>
<td>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</td>
<td>Dennis Kraemer, Zurich</td>
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<tr>
<td>17.00</td>
<td>Oral 10</td>
<td>Long-range PCR-based NGS applications to diagnose Mendelian retinal diseases</td>
<td>Jordi Maggi, Zurich</td>
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<tr>
<td>17.15</td>
<td>Oral 11</td>
<td>Investigating the role of the Bardet-Biedl protein Bbs1 in zebrafish using -omics approaches</td>
<td>Markus Masek, Zurich</td>
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<tr>
<td>17.30</td>
<td>Oral 12</td>
<td>Differentially expressed genes reflect disease-induced rather than disease-causing changes in the transcriptome</td>
<td>Eleonora Porcu, Lausanne</td>
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<tr>
<td>17.45</td>
<td>Oral 13</td>
<td>CoverageMaster: a clinical grade and user oriented CNV caller</td>
<td>Melivoia Rapti, Lausanne</td>
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</table>
| 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 |