Rare Diseases: From mechanisms to therapy and beyond

Program

Scientific Coordination

Arthur H M Burghes (Columbus, US)
Brunhilde Wirth (Cologne, DE) and Guenter Schwarz (Cologne, DE)

Cologne Ernst Klenk Symposium Committee 2019

Brunhilde Wirth - Guenter Schwarz (Chairs)
Oliver Cornely - Jörg Dötsch - Roman-Ulrich Müller – Christian Schaaf - Bernhard Schermer - Michal-Ruth Schweiger - Debora Grosskopf-Kroiher - Thomas Benzing

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Organized by
Center for Molecular Medicine Cologne (CMMC) - University of Cologne | University Hospital Cologne
www.cmmc-uni-koeln.de

Participation in the meeting is free of charge - free registration required.
Further information and free registration:
https://www.cmmc-uni-koeln.de/events/ernst-klenk-symposium/ernst-klenk-symposium-2019/

Sunday - Sept. 15, 2019

12.45 p.m. **Welcome Address**

Bettina Rockenbach Vice-Rector for Research and Innovation, University Cologne

Thomas Benzing Chair - Center for Molecular Medicine Cologne, University of Cologne

Eva Luise Köhler Chair - Board of Trustees of the Eva Luise & Horst Köhler Foundation and patroness of the *Allianz Chronischer Seltener Erkrankungen (Achse)*

Arthur H M Burghes - Brunhilde Wirth - Guenter Schwarz
Scientific Coordinators - Ernst Klenk Symposium 2019

Session I — State of the art in rare disease discovery

Chair Christian Schaaf (Inst. for Human Genetics)

1.00 - 1.30 p.m. Neurometabolic disease discoveries: Translating big data into better outcomes
Clara D M van Karnebeek - Academic Medical Centre, Univ. of Amsterdam - Amsterdam NL

1.30 - 2.00 p.m. The NIH undiagnosed diseases program: Expansion to a national and international network
William A Gahl - National Human Genome Research Institute - Bethesda, US

2.00 - 2.30 p.m. Genetic newborn population screening: Are we ready yet?
Georg F Hoffmann - University Children's Hospital - Heidelberg, DE

2.30 - 3.00 p.m. *Coffee Break*

Session II — Technologies to understand functional impact of human variation causing rare diseases

Chair Bernhard Schermer (Dept. II of Internal Medicine and CMMC)

3.00 - 3.30 p.m. Understanding the functional effects of coding variation at scale
Lea M Starita - Univ. of Washington and Brotman Baty Inst. for Precision Medicine - Seattle, US

3.30 - 4.00 p.m. From stem cells to organoids to disease modeling
Joseph V Bonventre - Harvard Insti. of Medicine, Brigham and Women's Hospital - Boston, US

4.00 - 4.30 p.m. Metabolomics - biomarker analysis as a functional read-out
Karlien L M Coene - Radboud Univ. Medical Center - Nijmegen, NL

4.30 - 5.00 p.m. *Coffee Break*

5.00 - 8.30 p.m. Poster session during the Welcome Get-Together (free finger food and drinks)
Selection of three posters for the Poster Awards

We are pleased to announce that we received 58 poster abstracts related to the following topics:

A 01 – A 17	Technologies to understand functional impact of human variation causing rare diseases
B 01 – B 08	Small molecule based therapy in neurological disorders
C 01 – C 03	Gene therapy to treat rare disorders
D 01 – D 08	Rare kidney and endocrine disorders and therapy
E 01 – E 12	Cancer and rare immune-mediated disease therapy and regulation
F 01 – F 06	Metabolic disorders and therapy
G 01 – G 04	Muscle, bone and dermis genetic disorders - from mechanism to treatment

Poster Award

The Klenk Poster Award 2019 of the Center for Molecular Medicine Cologne (CMMC) will be awarded to the presenting authors of the three most outstanding poster contributions. The awardees of the poster prizes are invited to give a short presentation (10 min.) during the last session on Tuesday, Sept. 17, 2019.

Monday - Sept. 16, 2019

Session III — Small molecule-based therapy in neurological disorders

Chair: Brunhilde Wirth (Inst. for Human Genetics)

9.00 - 9.30 a.m. **Antisense oligonucleotide based therapies for neurodegenerative diseases**
Frank C Bennett - Department of Research Biology, Ionis Pharmaceuticals - Carlsbad, US

9.30 - 10.00 a.m. **Rescuing a fatal disease, Spinal Muscular Atrophy, by antisense oligonucleotide therapy**
Richard S Finkel - Division of Pediatric Neurology, Nemours Children's Hospital - Orlando, US

10.00 - 10.15 a.m. *Coffee Break*

10.15 - 10.45 a.m. **A genome-wide suppressor paradigm for drug discovery in ciliopathies**
Nicholas Katsanis - Center for Human Disease Modeling, Duke University - Durham, US

10.45 – 11.15 a.m. **Antisense splicing modulation for familial dysautonomia**
Adrian R Krainer - Cold Spring Harbor Laboratory - Cold Spring Harbor, US

11.15 - 11.30 a.m. *Coffee Break*

Session IV — Gene therapy to treat rare disorders

Chair: Arthur M Burghes (The Ohio State University Wexner Medical Center, Columbus US)

11.30 - 12.00 p.m. **AAV-mediated gene therapy in neuromuscular disease: clinical immunology considerations**
Barry J Byrne - Powell Gene Therapy Center, Dept. of Pediatrics, Univ. of Florida - Gainesville, US

12.00 - 12.30 p.m. **Recent advances in gene therapy for Friedreich ataxia**
Hélène Puccio – Inst. of Genetics and Molecular and Cellular Biology (IGBMC) - Illkirch, FR

12.30 - 2.00 p.m. *Lunch Break with Poster Presentation (free soup for all participants)*

Session V — Rare kidney and endocrine disorders and therapy

Chair: Roman-Ulrich Müller (Dept. II of Internal Medicine)

2.00 - 2.30 p.m. **Molecular challenges in the treatment of complex genetic endocrine defects**
Heiko Krude – Inst. of Experimental Pediatric Endocrinology, Charité Berlin - Berlin, DE

2.30 - 3.00 p.m. **Current and potential treatments for ADPKD targeting cyclic AMP signaling**
Vicente E Torres - Mayo Clinic Translational Polycystic Kidney Disease Center - Rochester, US

3.00 - 3.30 p.m. **Membranous nephropathy: From molecules to disease and therapy**
Pierre Ronco - French National Institute of Health and Medical Research (INSERM - Paris, FR

3.30 - 3.50 p.m. *Coffee Break*

3.50 - 4.00 p.m. **Announcement of the Poster Awards** - Brunhilde Wirth and Guenter Schwarz

Session VI — Cancer and rare immune-mediated disease therapy and regulation

Chairs: Michal-Ruth Schweiger (Translational Epigenetics and Tumor Genetics) and Oliver Cornely (Clinical Trials Center Cologne)

4.00 - 4.30 p.m. **Epigenetic signatures in the blood of patients with cancer**
Michael R Speicher - Institute of Human Genetics, Medical University of Graz - Graz, AT

4.30 - 5.00 p.m. **TTP: Pathogenesis and the therapeutic path**
Marie Scully - Dept. of Haematology, University College London Hospital - London, GB

5.00 - 5.30 p.m. *Coffee Break*

5.30 - 6.30 p.m. **Ernst Klenk Lecture**
Laudation: Brunhilde Wirth
Spinal Muscular Atrophy from gene to treatment and the future
Arthur H M Burghes - The Ohio State University Wexner Medical Center - Columbus, US

Tuesday - Sept. 17, 2019

Session VII — Metabolic disorders and therapy

Chair: Guenter Schwarz (Inst. for Biochemistry)

9.00 - 9.30 a.m. Complex pathogenic cascades in lysosomal disorders and their implications for therapy
Frances Platt - Dept. of Pharmacology, University of Oxford - Oxford, GB

9.30 - 10.00 a.m. Therapeutic enhancement of autophagy for the treatment of lysosomal storage disorders
Carmine Settembre - Telethon Institute of Genetics and Medicine - Pozzuoli-Napoli, IT

10.00 - 10.30 a.m. Short presentation
given by the three poster awardees (3 x 10 min. short talk)

10.30 - 11.00 a.m. *Coffee Break*

Session VIII — Muscle, bone and dermis genetic disorders - from mechanism to treatment

Chair: Jörg Dötsch (Dept. of Pediatrics)

11.00 - 11.30 a.m. Novel treatments for osteogenesis imperfecta
Frank Rauch - Shriners Hospital for Children – Canada, McGill University - Montreal, CA

11.30 - 12.00 p.m. Sweating matters: Can protein replacement in utero correct anhidrotic ectodermal dysplasia?
Holm Schneider - Center for Ectodermal Dysplasias, Univ. Hospital Erlangen - Erlangen, DE

Session IX — What's next?

12.00 - 12.30 p.m. Affordable diagnosis and prevention of genetic disease
Sir John Burn - Institute of Genetic Medicine, Newcastle University - Newcastle upon Tyne, GB

12.30 - 12:45 p.m. Summary, open questions, concluding remarks
Arthur H M Burghes - Brunhilde Wirth - Guenter Schwarz

General Information

The meeting is open and no registration or attendance fee is required.
Participation is free of charge - free registration is required.

Childcare is provided by Spielland, University Hospital Cologne.

If you require an official invitation letter for travel refunding and visa issues or childcare, please contact:

Debora Grosskopf-Kroiher - debora.grosskopf-kroiher@uni-koeln.de

Center for Molecular Medicine Cologne (CMMC), Univ. of Cologne
CMMC Research Building, Robert-Koch-Str. 21, 50931 Cologne, Germany

Venue

Main Lecture Hall, Faculty of Medicine, University of Cologne
Access only possible from Joseph-Stelzmann-Str. 26 - please follow the yellow Klenk signs.
https://www.cmmc-uni-koeln.de/events/ernst-klenk-symposium/ernst-klenk-symposium-2019/venue/

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