



Eva Luise und Horst Köhler Stiftung



International Symposium Rare Disease Day 2016

Organized by

Charité Berlin Center for Rare Diseases
Berlin Institute of Health
Eva Luise und Horst Köhler Stiftung

in collaboration with

the Berlin Brandenburg Academy of Sciences (BBAW)
with support of Pfizer Pharma GmbH

Date: 03.03.2016 - 04.03.2016

Venue: Berlin Brandenburgische Akademie der Wissenschaften
Jägerstraße 22/23
10117 Berlin
www.bbaw.de

**Thursday,
8.30 – 9.00**

03. March 2016

**Welcome
by the BIH Director
and the Collaborative Research
Group on Rare Congenital Diseases**

Erwin Böttinger
Carmen Birchmeier
Christian Rosenmund

9.00 – 9.30

**Key note lecture :
Rare diseases - a key to precision medicine**

Veronica van Heynigen,
London

9.30 – 10.00

**Common pathways and transcription
network control in intellectual disability
and microcephaly**

Angela Kaindl (Charité)
Christian Rosenmund
(Charité)

10.00 – 10.30

**Towards a better understanding of
congenital endocrine diseases**

Carmen Birchmeier (MDC)
Heiko Krude (Charité)

10.30 – 11.00

Coffee break

11.00 – 11.30

**Mis-regulated chromatin folding as a cause
of congenital disease**

Stefan Mundlos (Charité)
Ana Pombo (MDC)

11.30 – 12.00

**Integrative omics-based dissection of
molecular mechanisms underlying
congenital abnormalities of the kidney and
the urinary tract**

Wei Chen (MDC)
Dominik Muller (Charité)

12.00 – 12.30

**Transcription network controlling heart
development and congenital heart disease**

Uwe Ohler (MDC)
Silke Rickert-Sperling
(Charité)

12.30 – 13.00

General Discussion

13.00 – 14.00

Lunch

14.00 – 14.15

Welcome adress

Annette Grüters-Kieslich
(Charité)
Martin Groetschel (BBAW)

14.15 – 14.45

**Keynote Lecture
Cellular and molecular basis of heart
development and function**

Prof.Rolf Bodmer,
BIH Fellow, La Jolla/Berlin

14.45 – 16.00

**Round table I
(Moderator: A. Grüters)**

O.Hiort (ERN)
H. Jueppner (Harvard,USA)
M.Dattani (UCL, London,
UK)

**Centers for Rare Diseases: International
Experiences**

16.00 – 17.30

**Round Table II
The German Healthcare System- Ready for
Rare Diseases?
(Moderator: G. Hoffmann)**

Pharma (P.Loeschmann)
Health Insurance
(D.Parow)
Hospital Society
(M. Schreiner),
MFT (F. Wissing),
Achse (C. Mundlos)

17.30 – 18.15

**Overlapping phenotypes of G-Protein
disorders**

Harald Jueppner,
Harvard Medical School,
Boston

18.30 – 21.00

Get together



Friday,

04. March 2015

9.00 – 9.05

Welcome

Peter Loeschmann, Pfizer

9.05 – 9.30

How to diagnose a rare disease

Stefan Mundlos (Charité)

9.30 – 11.10

**Advances made in Rare Disease Research:
Presentations of former Eva Luise Köhler
Research Award Recipients**

1. Lysosomal Storage Disease
2. Hypokalemic paralysis
3. Sickle cell anemia
4. Human iPSC cell model of 3 long QT syndrome

Volkmar Gieselmann, Bonn
Andreas Pflugrad, Ulm
Stefan Lobitz, Berlin,
Boris, Greber, Münster

11.10– 11.30

Coffee break

11.30 – 13.00

**Three Case Studies:
Segmental overgrowth syndrome
Interferonopathies
Hypothalamic disorders**

Jochen Rößler, Freiburg
Gerd Horneff, St Augustin
Mehul Dattani, GOSH,
London

13.00 – 14.00

Lunch

14.00 – 15.40

**Advances made in Rare Disease Research:
Presentations of former Eva Luise Köhler
Research Award Recipients**

5. Osteogenesis imperfecta
6. Primary ciliary dyskinesia
7. ALS
8. Cell therapy of pulmonary diseases
9. Wiskott-Aldrich Syndrome

Oliver Semler, Köln
Heymut Omeran, Münster
Niels Birbaumer, Tübingen
Gesine Hansen, Hannover
Christopher Baum,
Hannover

15.45 – 16.15

Coffee break

16.15 – 17.30

**Three Case Studies
Chorea Huntington
TTR Amyloidosis – Mutations, Phenotypes and
Epigenetics
Juvenile Obesity**

Katrin Bürk, Marburg

Anna Barreiros, Regensburg
Peter Kühnen, Berlin

17.30 – 18:30

**Key note lecture
Differences in sexual differentiation: Rare, but
no disease**

John Acherman, UCL Great
Ormond Street Hospital,
London

Saturday

05. March 2015

11.00 – 14.00

**Festliche Matinée
Eva Luise Köhler Forschungspreis 2016
Konzerthaus am Gendarmenmarkt
mit
Cellist Jan Vogler und
der Staatskapelle Halle
unter der Leitung von Maestro
Josep Caballé Domenech**