

22 -24 February 2012
Freiburg, Germany

International Meeting on

RARE DISEASES - Mechanisms and New Therapeutic Approaches

Wednesday, February 22

- 18:00 Interdisciplinary Session –
FCRD Presents its Activities
20:00 Get-Together

Thursday, February 23

- 13:00 **Opening Ceremony**
Session 1 **New Technologies – New Disease Causes**
13:15 Friedhelm Hildebrandt
*Whole Exome Sequencing Reveals
Mechanisms of Rare Childhood Diseases*
13:50 Matt Warman
*Does Increasing Bone Mass also Increase Bone
Strength in Mouse Models of
Osteogenesis Imperfecta*
14:30 Bodo Grimbacher
*Mendelian Traits in the Susceptibility to
Fungal Infections*
15:00 Coffee Break and Poster Session 1
Session 2 **Molecular Diseases Mechanisms and
Therapies**
16:00 Eli Sprecher
*(Rare)²: The Genetics of Uncommon Skin
Disorders in Population Isolates*
16:40 Jakub Tolar
*Stem Cell Transplantation for the Therapy
of Severe Epidermolysis Bullosa*
17:20 Nurten Akarsu
*Molecular Genetics of
Popliteal Pterygium Syndrome*
17:40 Marcus Mall
*Cystic Fibrosis: Emerging Therapies Targeting
the Basis Defect*
18:00 **Special Lecture**
Norbert Paul
*Ethical Dimensions of Treating a Rare Case
of Pompe Disease*
19:00 **Conference Dinner**

Friday, February 24

- Session 3 **Gene and Protein Therapies**
08:30 Carsten Bönnemann
*New Therapeutic Strategies for Muscular
Dystrophies*
09:10 Toni Cathomen
*Targeted Genome Editing as a Novel Therapeutic
Approach to Tackle Immune Deficiencies*
09:50 Short Talks
10:00 Coffee Break
Session 4 **Molecular Diseases Mechanisms**
10:30 Christian Kratz
Monogenic and Complex Cancer Syndromes
11:10 Bernd Wollnik
*Beyond FGF-Signaling: Novel Molecular Findings
in Patients with Syndromic Craniosynostosis*
11:40 Short Talks
12:30 Lunch and Poster Session 2
Session 5 **Compensational Therapy Strategies**
14:00 William Horton
*Insights into the Molecular Pathogenesis
and Treatment of Achondroplasia*
14:40 Short Talks
16:00 Coffee Break
Session 6 **New Genes and Molecular Diseases
Mechanisms**
16:30 Stephan Ehl
*When the Immune System Runs Amok -
Hemophagocytic Lymphohistiocytosis*
17:10 Maximilian Muenke
*Bringing Rare Diseases to the Masses:
Why Understanding Orphan Disorders Matters
to (Almost) Everyone*
18:00 Informal Get-Together

For further information and abstract submission please visit our website

www.rarediseasescongress.net

The meeting is hosted by the Freiburg Center for Rare Diseases; organization in cooperation with kongress&kommunikation gGmbH

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