

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 in FRIAS Lounge

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): 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 Short Talks
18:00 **Special Lecture**
Norbert Paul
**Ethical Dimensions of Treating a Rare Case
of Pompe Disease**
19:00 **Conference Dinner** in "Historisches Kaufhaus"

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 Talk
10:00 Coffee Break
Session 4 **Molecular Diseases Mechanisms**
10:30 Christian Kratz
Monogenic and Complex Cancer
11:10 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

sponsored by