Presenting Author Titel

Freitag

8.30 h – 9.00 h Welcome / Bievenue / Begrüßung

9.00 h – 10.50 h Experimental therapies and new therapeutic concepts in preclinical models

Arsenijevic Derivation of Traceable Photoreceptors from Mouse Embryonic Stem Cells: implications for

Cell Therapy and Drug Screening.

Ader Restoration of photopic responses following cone-like photoreceptor transplantation into a

cone degeneration mouse model

Seeliger Gene Therapy in Achromatopsia: From Preclinical Models to Therapeutic Application

Miranda de Sousa Gene Replacement Therapy for CSNB as A Proof-Of-Principal for Other Inner Retinal

Disorders

Yanik Highly Specific Nucleases to target the RPGR Gene for Gene Therapy

Bonifert Antisense Oligonucleotide Mediated Rescue of a Deep Intronic Point Mutation in OPA1

Nagel-Wolfrum Ignore the stop: translational read-through of nonsense mutations in Usher syndrome and

related ciliopathy genes

Frohns Inefficient repair of DNA double-strand breaks in murine rod photoreceptors

Jagodzinska Wfs1-/- mice: phenotyping and gene therapy against Wolfram Syndrome disease

10.50 – 11.20 Coffee

11.20 – 12.05 h Key Note Lecture

Hamel The RPE65 story: from discovery to treatment of patients

12.10 h – 13.00 h Business Meetings

13.00 h – 14.00 h Lunch

14.00 h- 14.50 h Targeting neuronal cell death for the treatment of hereditary retinal degeneration

Paquet-Durand A better understanding of cell death to guide the rational development of new therapeutic

approaches for hereditary retinal degeneration.

Mühlfriedel A key role for cyclic nucleotide gated (CNG)channels in cGMP-related retinitis pigmentosa:

Photoreceptor survival in Cngb1-/- x rd1 double mutants

Grosche How Müller glia affect neuronal survial after transient ischemia

Müller, B. Time course of gene expression in murine retinal explant cultures

14.50h – 15.35 h Key Note Lecture

Reichenbach Retinal Glial Cells - Why they are of Interest to Ophthalmologists

15.40 h – 16.10 h Coffee

16.10 h -18.30 h Cases you do not see every day

Munier NBAS-related Retinal Dystrophy

Alex Unklares Makulaödem bei einem 9-jährigen Jungen

Preising Genotype-Phenotype correlations in gene mutation carriers of PRPF31 Mutations

Escher Atypical mild Goldmann-Favre syndrome with residual rod function in presence of

homodimerization-competent NR2E3 mutant proteins

Vaclavik Cone dystrophy and sterility – follow up

Vaclavik New case of Malatia Leventinese with a de novo mutation Solbach A Bull's eye fundus autofluorescence points towards EYS

Hamel An unusually severe retinal dystrophy in a child

Besgen Girl with cornea verticillata

Wenner Case of a patient with retinal dystrophy and neurologic symptoms

Leroy Variable Phenotype and Retinal Abnormalities in Ectopia Lentis et Pupillae

Cosendai Argus II Retinal Prosthesis System: Results from the European Post-Market Surveillance

Study

18.30 h - 19.30 h Vines and Spirit

20.00 h Dinner at Heyligenstaedt

Samstag

8.30 h - 9.00 h Approaching disease function

> Orhan Genotypic and Phenotypic Characterization of line 1-P23H rat model Karlstetter The Fam161a-Gene trap mouse as novel ciliopathy mouse model

Wolfrum Decoding of protein networks reveals insights in the molecular basis of the Usher

syndrome and related ciliopathies

9.00 h - 10.30 h Natural history of disease as basis for the detection of therapeutic effects

> Kellner Non-invasive retinal imaging: The primary tool for the diagnosis of inherited retinal

Gliem Evaluation of Quantitative Fundusautofluorescence in Healthy Controls and Stargardt's

Disease

Monoallelic Mutations in ABCA4 Are Not Associated With Abnormal Lipofuscin

Accumulation

Neuille LRIT3 is essential for the proper membrane localization of different components of the ON-

bipolar cell signaling cascade

Lenaers An update on the Genetics of Inherited Optic Neuropathies

Matet Multimodal imaging using quantitative autofluorescence in achromatopsia: a case series Schorderet Characterization of HMX1, the gene responsible for the Schorderet-Munier-Franceschetti

auriculo-ocular syndrome.

10.30 h - 11.00 h Coffee

Müller, P.

11.00 h – 12.20 h New developments in genetics or clinical diagnosis

> Lorenz Two-color-pupillometry

Hauck Unbiased allele-specific quantitative proteomics unravels molecular mechanisms

influenced by cis-regulatory genomic variations

Copy Number Variation detection from Next Generation Sequencing data in inherited Audo

retinal disorders

Zanlonghi Whole exome sequencing in a multiplex family case of Plateau Iris Syndrome

The importance of Sanger sequencing of RPGR exon ORF15 in the time of next-generation Zeitz

sequencing

Bolz Comprehensive genetic analysis of Usher syndrome by next-generation sequencing Identification of mutations in retinal disease genes using Whole Exome Sequencing Mayer

12.20 h Farewell / Archèvement / Verabschiedung