SCHEDULE

Thursday 27th

09.00 – 09.15 Opening

Session 1: Therapy, today and tomorrow

Chair: William J. Kimberling

09.15 – 09.45 Paul A. Sieving. National Eye Institute. Bethesda, MD, USA.
Clinical trials of CNTF for retinitis pigmentosa

09.45 – 10.15 Nicolás Cuenca. Departamento de Fisiología, Genética y Microbiología, Universidad de Alicante. Alicante, Spain.
Neuroprotective effect of antiapoptotics and antioxidants as a therapeutic option for retinitis pigmentosa

10.15 – 10.45 Tamar Ben-Yosef. Rapaport Faculty of Medicine, Technion-Israel Institute of Technology. Haifa, Israel.
The use of aminoglycosides and their derivatives as a therapy for Usher syndrome

10.45 – 11.15 David S. Williams. Jules Stein Eye Institute, UCLA School of Medicine. Los Angeles, CA, USA.
Gene therapy for Usher syndrome type 1B

11.15 – 11.45 Coffee break

11.45 – 12.15 Kerstin Nagel-Wolfrum. Department of Cell and Matriz Biology, Johannes Gutenberg-University of Mainz. Mainz, Germany.
USH1C therapy strategies in the retina

12.15 – 12.45 Peter Francis. Degeneration Center, Casey Eye Institute. Oregon, USA.

Stem cell therapy for Usher 2a

12.45 – 13.15 Eeva-Marja Sankila. Department of Ophthalmology, University of Helsinki, Finland.

Gene therapy for Usher syndrome type 3

13.15 – 14.45 Lunch

Session 2: Psychosocial aspects

Chair: Claes Möller

14.45 – 15.15 Claes Möller. Department of Audiology, Örebro University Hospital. Örebro, Sweden.

State of the art clinical and genetic diagnosis and early intervention in Usher syndrome

15.15 – 15.45 Ilene D. Miner. LCSW Private Practice. Venice, CA, USA.

Psychosocial impact of Usher syndrome: Adults and the family

15.45 – 16.15 Coffee break

16.15 – 16.45 Berth Danermark. The Swedish Institute for Disablility Research, Örebro University. Örebro, Sweden.

Usher syndrome and psychosocial health

16.45 - 17.45 Oral Session

E. van Wijk

Th USH2A Interaction Partner NINL isoB Associates with BBS6, plays a role in establishing planar cell polarity and functions in cilia assembly

Shzeena Dad

Identification of a new Usher 3 like locus

Ole E. Mortensen
How do people with Usher Syndrome Live their lives?

M. Westerfield
Usher scaffold proteins provide complementary functions in retina and inner ear

Berth Danermark
Deafblindness and the notion of trust, ontological security, social recognition and self-identity

Friday 28th

Session 3: Natural history studies
Chair: Eduardo Duarte Silva

09.00 – 09.30 Maria Bitner-Glindzicz. Institute of Child Health. London, UK.
Lessons from the UK National Collaborative Usher Study

09.30 – 10.00 Sten Andréasson. Department of Ophthalmology, University of Lund. Lund, Sweden.
Development of the ERG in the first five years of life

10.00 – 10.30 Samuel G. Jacobson. Scheie Eye Institute, University of Pennsylvania. Philadelphia, USA.
Retinal Disease Expression in the Usher syndrome

10.30 – 11.00 Margaret A. Kenna. Department of Otolaryngology, Chilrden's Hospital. Boston, MA, USA.
Vestibular function in children with Usher syndrome: What do we know and how should we study it?

11.00 – 11.30 Coffee break

Session 4: Population genetics and epidemiology
Chair: Margaret A. Kenna

11.30 – 12.00 William J. Kimberling. Departments of Ophthalmology and Visual Sciences and Otolaryngology, University of Iowa Carver School of Medicine. Iowa City, USA and Boys Town National Hospital. Omaha, USA.

Screening and early diagnosis

12.00 – 12.30 Anne-Françoise Roux. Institut Universitaire de Recherche Clinique. Montpellier, France.

The utility of databases in diagnosis

12.30 – 13.00 Carmen Ayuso. Fundación Jiménez Díaz, Clínica Ntra. Sra. de la Concepción. Madrid, Spain.

Epidemiology of Usher syndrome

13.00 – 13.30 Richard JH Smith. Department of Otolaringology, University of Iowa. Iowa, USA.

Developing more comprehensive genetic screening strategies for congenital sensorineural hearing loss

13.30 – 15.00 Lunch

Session 5: Development of Animal models

Chair: Peter Francis

15.00 – 15.30 Nicholas Katsanis. Duke University Medical Center. Durham, NC, USA.

Modifiers of ciliary disease

15.30 – 16.00 Ray Iezzi. Department of Ophthalmology, Vitreoretinal Service, College of Medicine, Mayo Clinic. Rochester, MN, USA.

Rat models of retinal degenerations: therapeutic interventions and clinical correlations. I

16.00 – 16.30 Coffee break

16.30 – 17.00 Arlene Drack. Department of Ophthalmology and Visual Sciences, University of Iowa. Iowa, USA.

Mouse models of retinal degenerations: therapeutic

interventions and clinical correlations. II

17.00 – 17.30 Isabel Varela-Nieto. Group of Neurobiology of Hearing, Institute for Biomedical Research CSIC-UAM. Madrid. Spain.
Non-invasive evaluation of hearing in mouse models of deafness: a focus on IGF-I deficiency

Saturday 29th

Session 6: Molecular and biochemical aspects
Chair: Roser González-Duarte

09.00 – 09.30 Hanno J. Bolz. Institute of Human Genetics, University Hospital of Cologne. Köln, Germany.
New strategies/technologies to identify new genes

09.30 – 10.00 Hannie Kremer. Radboud University Nijmegen Medical Centre. Nijmegen, The Netherlands.
The Usher protein network in the inner ear

10.00 – 10.30 Uwe Wolfrum. Department of Cell Biology, The Scripps Research Institute. La Jolla, CA, USA.
The Usher protein network in the retina

10.30 – 11.00 Coffee break

Session 7: Diagnostic
Chair: José M. Millán

11.00 – 11.30 Bernhard H. F. Weber. Institute of Human Genetics, University of Regensburg. Regensburg, Germany.
RetChip1.0 – A novel array-based tool for diagnostic testing in hereditary retinal degenerations

11.30 – 12.00 Roser González-Duarte. Departament de Genètica. Universitat de Barcelona. Barcelona, Spain.
Challenges of the genetic diagnosis of highly heterogeneous disorders

12.00 – 12.30 Heidi L. Rehm. Laboratory for Molecular Medicine, Partners Healthcare Center for Personalized Genetic Medicine. Boston, MA, USA.
The Otochip sequencing array for hearing loss and Usher

12.30 – 13.00 Ilona Lind. Asper Biotech. Tartu, Estonia.
Asper's diagnostic tool for the Usher syndrome