



4th European Days of Albinism, 7-11 March 2018

Hurdal Vision and Activity Center, Oslo, Norway

Organized by Albinism Europe and NFFA



ALBINISM EUROPE



SCIENTIFIC and SOCIAL PROGRAM

Wednesday, 7 March 2018

Airport shuttle to Hurdalsenteret and Check in

18:00-20:00 Welcome dinner at Hurdalsenteret

Thursday, 8 March 2018

7:30- 8:30 *Breakfast*

8:45- 9:30 **WELCOME SESSION**

Edel Krøll (4th EDA Coordinator for NFFA)

Kjeld Christian Fjeldvig (President of NFFA)

Antoine Gliksohn (Albinism Europe Coordinator)

Benoît Arveiler & Lluís Montoliu (Albinism Europe Scientific Committee co-Chairs)

9:30-10:50 (I) **FIRST SCIENTIFIC SESSION** Chair: Benoît Arveiler

9:30- 9:45 Setting the stage: EDAs and Albinism

Lluís Montoliu (Madrid, Spain) and **Benoît Arveiler** (Bordeaux, France)

9:45-10:10 Albinism: what's in a name?

Charlotte Kruijt and **Maria Van Genderen** (Zeist, Netherlands)

10:10-10:35 Molecular characterization of a series of 990 index patients with albinism

Benoît Arveiler (Bordeaux, France)

10:35-10:50 Discussion

10:50-11:10 *Coffee Break*

- 11:10-12:30 (II) **SECOND SCIENTIFIC SESSION** Chair: Alessandra del Longo
- 11:10-11:25 Multidisciplinary approach of Italian Albino patients
Lucia Mauri and **Alessandra del Longo** (Milan, Italy)
- 11:25-11:45 Lessons of a day hospital: comprehensive assessment of patients with albinism in a European setting
Fanny Morice-Picard (Bordeaux, France)
- 11:45-12:00 Albinism in Denmark
Karen Grønskov (Copenhagen, Denmark)
- 12:00-12:15 Burden of Albinism: creation of a questionnaire
Charles Taieb (Paris, France)
- 12:15-12:30 Discussion
- 12:30-13:30 *Lunch*
- 13:30-15:30 (III) **THIRD SCIENTIFIC SESSION** Chair: Lluís Montoliu
- 13:30-13:45 Clinical and genetic study of albinism in Russia
Vitaly Kadyshev (Moscow, Russia)
- 13:45-14:00 Albinism in Israel: clinical and genetic characteristics
Anat Blumenfeld (Jerusalem, Israel)
- 14:00-14:15 Albinism in Brazil
Carolina Marçon (São Paulo, Brazil)
- 14:15-14:30 Albinism in Spain
Lluís Montoliu (Madrid, Spain)
- 14:30-14:45 Who has got albinism?
Isabelle Drumare (Lille, France)
- 14:45-15:00 Attempt of genotype-phenotype correlation in a cohort of molecularly confirmed OCA1 patients presenting the R402Q variant associated with a Tyr pathogenic mutation
Catherine Duncombe-Poulet (Caen, France)
- 15:00-15:30 Discussion
- 15:30-16:00 *Coffee Break*
- 16:00-18:00 (IV) **FOURTH SCIENTIFIC SESSION** Chair: Michael Hoffmann
- 16:00-16:25 Factors limiting vision in albinism and prediction of visual function later in life
Irene Gottlob (Leicester, United Kingdom)
- 16:25-16:40 Altered organization of the visual cortex in FHONDA syndrome
Khazar Ahmadi (Magdeburg, Germany)

- 16:40-17:05 Albinism. Perceiving the visual world with an alternative visual system
Michael Hoffmann (Magdeburg, Germany)
- 17:05-17:20 Albinism in Israel: clinical characteristics
Claudia Yahalom (Jerusalem, Israel)
- 17:20-17:35 Ophthalmologic and psychophysical evaluation of the visual system of individuals with albinism
Ronaldo Sano (São Paulo, Brazil)
- 17:35-17:45 Interest of OCT in nystagmus exploration: two case-reports of a late diagnosis of albinism,
Dominique Brémond-Gignac (Paris, France)
- 17:45-18:00 Discussion
- 18:00-19:30 *Dinner*
- 20:00-22:00 **FREE EVENING**
- 21:00 *Refreshments, coffee, cakes*

Friday, 9 March 2018

- 7:30- 8:30 *Breakfast*
- 9:00-10:30 (V) **FIFTH SCIENTIFIC SESSION** Chair: Maria VAN GENDEREN
- 9:00- 9:25 Generation of new mouse models for investigating non-syndromic types of albinism through CRISPR gene-editing technology
Andrea Montero and **Lluís Montoliu** (Madrid, Spain)
- 9:25- 9:50 Variation in MFSD12 expression regulates skin pigmentation in African populations
Mickey S. Marks (Philadelphia, PA, USA)
- 9:50-10:15 Albinism in Africa: What can we learn from albinism frequency data?
Patricia Lund (Coventry, United Kingdom)
- 10:15-10:30 Discussion
- 10:30-11:00 *Coffee Break*
- 11:00-12:30 (VI) **SIXTH SCIENTIFIC SESSION** Chair: Fanny Morice-Picard
- 11:00-11:25 Reading in albinism
Frank A. Proudlock (Leicester, United Kingdom)
- 11:25-11:40 Analysis about visual acuity and binocular visual functions and the wavelet of children's nystagmus in albinism
Dayong Bai (Beijing, China)
- 11:40-12:00 Mild syndromic forms of albinism: HPS3, HPS5, HPS6 and HPS8
Perrine Pennamen (Bordeaux, France)

- 12:00-12:15 Observations of two unrelated children with OCA phenotype resulting in HPS1 mutations
Dominique Brémond-Gignac (Paris, France)
- 12:15-12:30 Discussion
- 12:30-13:30 *Lunch*
- 13:30-15:00 Special Social Activity – *Outdoor winter activities*
- 15:00-15:30 *Coffee Break*
- 15:30-17:30 (VII) **SEVENTH SCIENTIFIC SESSION** Chair: Irene Gottlob
- 15:30-16:00 Oral Nitisinone in Oculocutaneous Albinism, type 1b (OCA-1b): Results of a One-year Pilot Treatment Trial
Brian Brooks (Bethesda, MA, USA)
- 16:00-16:15 Discussion
- 16:15-16:45 Consensus guidelines for the diagnosis and management of albinism
Mervyn Thomas and **Irene Gottlob** (Leicester, United Kingdom)
- 16:45-17:30 Discussion
- 17:30-17:45 Building of therapeutic education program for albinism: a multidisciplinary partnership
Smail Hadj-Rabia (Paris, France)
- 19:00-20:00 *Dinner*
- 20:00-22:00 **SOCIAL EVENING**
- 20:00-20:30 Official group photo by the conference photographer (in the gym)
- 21:00-21:30 Short film projection “*Change your heart not my color*” followed by discussion

Saturday, 10 March 2018

- 9:00-11:45 **JOINT SESSION**
- 9:00-10:00 Albinism and Human Rights
Ikponwosa Ero (Nigeria, Canada) – UN Independent Expert on the enjoyment of Human Rights by persons with albinism
- 10:00-10:30 Coffee break
- 10:30-10:50 Update on the activities of Albinism Europe
Antoine Gliksohn (Paris, France)
- 10:50-11:00 Thoughts from my 20 years serving the Albinism community
Mike McGowan - NOAH (Chicago, USA)
- 11:00-11:30 Summary of scientific presentations
Albinism Europe Scientific Committee

- 11:30-11:45 Discussion
- 11:45-12:00 **CLOSING REMARKS**
- 12:00-13:00 *Lunch*
- 13:00 Shuttle bus to Oslo International Airport (for people leaving on Saturday March 10th)
- 13:00-18:00 FREE AFTERNOON – optional outdoor winter activities
- 18:30-20:00 Dinner
- 20:00-20:30 Short film projection “Moonlight Princess” followed by discussion with Nadine Nourney (Berlin, Germany), main actress and YPA participant
- 21:00 Refreshments, snacks

OPTIONAL - Sunday, 11 March 2018

- 7:30- 9:30 *Breakfast*
- 8:30-10:15 FREE MORNING - optional outdoor winter activities
- 12:30-13:30 *Lunch*
- 13:30 Shuttle bus to Oslo International Airport

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