4th European Days of Albinism, 7-11 March 2018
Hurdal Vision and Activity Center, Oslo, Norway
Organized by Albinism Europe and NFFA

SCIENTIFIC and SOCIAL PROGRAM

Wednesday, 7 March 2018

-17:00 SHUTTLE BUS FROM OSLO INTL AIRPORT TO HURDALSENTERET
18:00-20:00 WELCOME DINNER AT HURDALSENTERET

Thursday, 8 March 2018

07:30-08:30 Breakfast
08:45-09:30 WELCOME SESSION (Organizers: NFFA, Albinism Europe)
09:30-10:50 (I) FIRST SCIENTIFIC SESSION Chair: Benoît Arveiler
09:30-09:45 Setting the stage: EDAs and Albinism
Lluis Montoliu (Madrid, Spain) and Benoît Arveiler (Bordeaux, France)
09:45-10:10 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 (Fontenay-sous-Bois, France)

12:15-12:30 Discussion

12:30-13:30 Lunch

13:30-15:30 (III) THIRD SCIENTIFIC SESSION Chair: Lluis Montoliu

13:30-13:45 Clinical and genetic study of albinism in Russia
Vitaly Kadyshhev (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
Lluis 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

18:00-19:00 Dinner

20:00-21:00 Special Evening Session

Friday, 9 March 2018

07:30-08:30 Breakfast

09:00-10:30 (V) FIFTH SCIENTIFIC SESSION Chair: Maria VAN GENDEREN

09:00-09:25 Generation of new mouse models for investigating non-syndromic types of albinism through CRISPR gene-editing technology
Andrea Montero and Lluis Montoliu (Madrid, Spain)

09:25-09:50 Variation in MFSD12 expression regulates skin pigmentation in African populations
Mickey S. Marks (Philadelphia, PA, USA)

09:50-10:15 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

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

19:00-20:30  Gala Dinner

21:00-22:30  Social Evening

**Saturday, 10 March 2018**

07:30-08:30  Breakfast

08:30-10:15  JOINT SESSION WITH PATIENTS' ASSOCIATIONS AND SPECIAL SPEAKERS

10:15-10:45  Coffee Break

10:45-12:00  4TH EDA CLOSING SESSION

12:00-13:00  Lunch

13:30-  FIRST SHUTTLE BUS TO OSLO INTERNATIONAL AIRPORT
OPTIONAL - Sunday, 11 March 2018

07:30-09:30  Breakfast
08:30-10:15  FREE MORNING (OPTIONAL OUTDOOR ACTIVITIES)
12:30-13:30  Lunch

With the financial support of the following organizations: