



## 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

- 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  
**Lluís 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: 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
- 18:00-19:00 *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 **Lluís 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*

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