

III European Days of Albinism (3rd EDA)
Milan, 7-8 April 2016

3rdEDA Scientific Committee:

Arveiler Benoît, (Bordeaux)
Del Longo Alessandra, (Milan)
Käsmann-Kellner Barbara, (Homburg/Saar)
Montoliu Lluis (Madrid)
Patrosso Maria Cristina (Milan)
Penco Silvana, (Milan)
Piozzi Elena, (Milan)
Primignani Paola, (Milan)
Schiaffino Maria Vittoria, (Milan)

Thursday, 7 April 2016

Plenary welcome session: scientists and associations together

08:15 -08:45 Registration (all together)

08:45-09:15 General welcome (all together)

Chair: *Tronconi E, Marocchi A, Gesu G, Representative of the Region, Masto R, Paschetta L*

Concurrent sessions: scientists and associations separated

09:15-10:15 Scientific Session I

Clinical Definition of Albinism

Chair: *Gottlob I, Piozzi E*

Expanding the phenotype of albinism? - Barbara Käsmann-Kellner (*20 min*)

Phenotypic spectrum in FHONDA and albinism- Maria Van Genderen (*20 min*)

Discussion (20 min)

10:15 -11.45 Scientific Session II

Clinical and Molecular Diagnosis of Albinism

Chair: *Gronskov K, Scalinci S*

Postnatal retinal development in albinism as compared to healthy controls – Irene Gottlob I- (*20 min*)

Visual cortex in albinism - anatomy and function – Michael Hoffmann (*20 min*)

Electrophysiological aspects in albino patients – Alma Patrizia Tormene P (*15 min*)

Molecular diagnostic approaches: Targeted sequencing, exome, array-CGH? - Benoit Arveiler B (*20 min*)

Discussion (15 min)

11:45-12:10 coffee break

12:10-13:30 Scientific Session III

European Diagnostic Experiences

Chair: Taïeb A, Primignani P

Clinical and molecular experience in Denmark- Karen Grønskov (*20 min*)

Clinical and molecular experience in Italy – Alessandra Del Longo, Lucia Mauri L (*15 min*)

Albino Day in Madrid- Marta Cortón (*15 min*)

Albino Day in Bordeaux – Fanny Maurice Picard (*15 min*)

Discussion (*15 min*)

13:30-14:40 Lunch break (all together)

**14:40- 16:00 Scientific Session IV:
Pathogenesis of Albinism**

Chair: *Montoliu L, Schiaffino MV,*

Neurogenesis in animal models – Alexandra Rebsam (*20 min*)

Syndromic albinism disorders affecting membrane trafficking during melanosome formation- Michael Marks (*20 min*)

Melosome PH – Elena Oancea (*20 min*)

Discussion (*20 min*)

16:00-16.30 Coffee/Tea break

**16:30-18:10 Scientific Session V:
Beyond Eyes**

Chair: *Arveiler B., Penco S.*

Oculocutaneous albinism in East and Southern Africa - Patricia Lund (*20 min*)

Skin photoprotection issues in albinism - Alain Taïeb (*20 min*)

Dermatological characterization in Italian non adults albinos – Donata Calò (*20 min*)

Audiological deficits in albinos: correlation with genetic/clinical phenotype – Giuseppe Mingoia (*20 min*)

Discussion (*20 min*)

Friday, 8 April 2016

**8.30-10:00 Scientific Session VI
Technical Support and Therapeutic Frontiers**

Chair: *Käsmann-Kellner B, Del Longo A*

Visual management training in low vision patients - Flavia Fabiani
(15 min)

Pharmacological therapy – Brian Brooks (20 min)

Gene Therapy – Enrico Surace E (15 min)

New animal models of albinism generated with CRISPR tools – Lluis Montoliu (20 min)

Discussion (20 min)

10.00 – 10.30 Final Scientific Discussion:

Chairs: *Schiaffino MV, Marocchi A, Gottlob I, Montoliu L*

Discussion (30 min)

Plenary final session: scientists and associations together

10.30 -11:00 Summary of scientific presentations (*Käsmann-Kellner B, Patrosso MC*)

11:00 - 11.20 Summary of psycho pedagogical presentations (20 min)

11.20-11.50 Summary of association's presentations (30 min)

11.50- 12.00 Kid for kindness presentation (10 min)

12.00- 12.10 Dermatological Prevention presentation (10 min)

12.10-13.30 Global discussion/Presentation of leaflet/book on albinism for the general public/Proposal for the next meeting.