



2nd European Days of Albinism (2EDA)

**5-6 April 2014, Venue: ONCE in Valencia
Gran Vía Ramón y Cajal 13, 46007 Valencia, Spain**

Organized by ALBA (www.albinismo.es), the Spanish association in support of people with albinism

2EDA Scientific Committee

Lluís Montoliu (Madrid, Spain)
Karen Grønskov (Copenhagen, Denmark)
Vittoria Schiaffino (Milano, Italy)
Barbara Käsmann-Kellner (Homburg, Germany)
Benoît Arveiler (Bordeaux, France)

Confirmed invited speakers

Robert Aquaron (Marseille, France)
Esteban Dell'Angelica (Los Angeles, CA, USA)
Benoît Arveiler (Bordeaux, France)
Murray Brilliant (Marshfield, WI, USA)
Carmen Carranza Romero (Madrid, Spain)
José Carlos García-Borrón (Murcia, Spain)
Maria van Genderen (Zeist, The Netherlands)
Michael Hoffmann (Magdeburg, Germany)
Barbara Käsmann-Kellner (Homburg, Germany)
Herbert Kirchesch (Pulheim, Germany)
Alessandra Del Longo (Milan, Italy)
Valeria Marigo (Modena, Italy)
Mónica Martínez-García (Madrid, Spain)
Lucia Mauri (Milan, Italy)
Lluís Montoliu (Madrid, Spain)
Shari Parker (Sydney, Australia)
Fanny Morice-Picard (Bordeaux, France)
Elena Piozzi (Milan, Italy)
Markus Preisig (Giessen, Germany)
Frank Proudlock (Leicester, UK)
Alexandra Rebsam (Paris, France)
Vittoria Schiaffino (Milan, Italy)
Enrico Surace (Naples, Italy)
Tamio Suzuki (Yamagata, Japan)
Alain Taïeb (Bordeaux, France)
Pedro de la Villa (Madrid, Spain)

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FINAL Program

Saturday, 5 April 2014 (concurrent sessions: scientists and associations separated)

08:30-09:00 **Registration**

09:00-09:45 **General welcome, ALBA, ONCE authorities, Organizers**
(all participants together: researchers, associations, youth with albinism)

Session 1: Human molecular genetics: genes associated with albinism
(Chair: Lluís Montoliu)

09:45-10:15 **Welcome and new genes associated with albinism** (Lluís Montoliu)

10:15-10:35 **OCA in Africa** (Robert Aquaron)

10:35-10:55 **FHONDA** (Maria Van Genderen)

10:55-11:30 Panel Discussion

11:30-12:00 Coffee/Tea break

Session 2: Human genetics session: molecular diagnosis of albinism
(Chair: Benoît Arveiler)

12:00-12:30 **Genetic diagnosis of albinism in France** (Benoît Arveiler)

12:30-12:50 **Genetic diagnosis of albinism in Spain** (Mónica Martínez-García)

12:50-13:10 **Genetic diagnosis of albinism in Italy** (Lucia Mauri)

13:10-13:40 **Genetic diagnosis of albinism in Japan** (Tamio Suzuki)

13:40-14:00 Panel Discussion

14:00-15:00 Lunch break

Session 3: Visual alterations associated with albinism
(Chair: Barbara Käsman-Kellner)

15:00-15:20 **Defining the ocular phenotype in albinism** (Barbara Käsman-Kellner)

15:20-15:35 **Ophthalmological examination of albino patients** (Elena Piozzi)

15:35-15:50 **Nystagmus in albinism** (Frank Proudlock)

15:50-16:05 **Chiasm misrouting-clinical perspective** (Michael Hoffmann)

16:05-16:20 **Chiasm misrouting-research perspective** (Alexandra Rebsam)

16:20-16:35 **Developmental implications of ocular features** (Markus Preising)

16:35-16:50 **Retinal electrophysiology in animal models** (Pedro de la Villa)

16:50-17:15 Panel Discussion

17:15-17:45 Coffee/Tea break

Session 4: Molecular and cell biology of albinism: Physiopathology of albinism

(Chair: Vittoria Schiaffino)

- 17:45-18:05 **Proteins involved in the synthesis of melanin** (José Carlos García-Borrón)
18:05-18:25 **Molecular pathogenesis of ocular albinism type I** (Vittoria Schiaffino)
18:25-18:45 **Syndromic albinism - Hermansky-Pudlak syndrome** (Esteban Dell'Angelica)
18:45-19:00 Panel Discussion

20:30 2EDA Conference Dinner

[Sunday, 6 April 2014 \(concurrent sessions: scientists and associations separated 09:00 – 11:30\)](#)

Session 5: Dermatological aspects of albinism

(Chair: Alain Taïeb)

- 9:00-9:15 **What's new in dermatology about albinism?** (Alain Taïeb)
9:15-9:30 **Dermatoclinical aspects of albinism in Europe** (Herbert Kirchesch)
9:30-9:45 **Dermatoclinical aspects of albinism in Africa** (Carmen Carranza)
9:45-10:00 Panel discussion

Session 6: Treatment of albinism, gene therapy, clinical trials

(Chair: Lluís Montoliu)

- 10:00-10:15 **Is treatment of albinism becoming a possibility?** (Lluís Montoliu)
10:15-10:30 **Clinical trials with L-DOPA** (Murray Brilliant)
10:30-10:45 **Stem cells and retinal cell regeneration** (Valeria Marigo)
10:45-11:00 **First gene therapy pre-clinical attempts** (Enrico Surace)
11:00-11:30 Panel discussion

11:30-12:00 Late coffee break/ light lunch

Combined final session: researchers and associations: Albino-days experiences

(Chairs: Lluís Montoliu and José Antonio Solves)

- 12:00-12:15 **Albino day in Bordeaux** (Fanny Morice-Picard)
12:15-12:30 **Albino day in Milano** (Alessandra Del Longo)
12:30-12:50 **Rehabilitation Medicine applied to albinism** (Shari Parker)

- 12:50-13:10 **Summary of scientific presentations** (Lluís Montoliu)
13:10-13:30 **Summary of associations presentations** (Florian Gliksohn)

13:30-14:00 **Global discussion / questions & answers**
(all scientists and association members)

14:30 **Lunch**

16:00 **End of the II EDA**

Departures