III EDA
3rd European Day of Albinism

Medical Section
Association Section
Psyco pedagogical Section

LOMBARDY REGION BUILDING
Piazza Città di Lombardia, Milan, Italy
ALBINIT with Niguarda Hospital organize, at the Lombardy Region Building in Milan, the Third European Days of Albinism (IIIEDA).

Meeting point for researchers focused on albinism, international associations of patients and their families to discuss both scientific and social aspects of this rare pathology, exchanging ideas, projects and initiatives.

A satellite meeting concerning the psycho-pedagogic aspects of this disease will be held simultaneously.

The meeting is divided into three sections:

- Room 5
  **Medical Section:**
  Physicians, Biologists and Researchers (only for operators in the albinism sector) - p.1
  ECM credits: 8.40

- Room 4
  **Associations Section:**
  only for representatives of Associations - p.6

- Room Biagi
  **Psyco pedagogical Section:**
  Orthoptists, Speech therapists, Laboratory technicians, social and health educators - p.8
  ECM credits: 4.90

*The Opening Ceremony and the Plenary final session are in the Room Biagi.*
MEDICAL SECTION

7/4/2016

8.00 - 8.30  Registration

8.30-9.00  Opening ceremony
Representative of the Region, G.P.Gesu, A.Marocchi, R.Masto, L.Paschetta, E.Tronconi

9.00-10.00  **SCIENTIFIC SESSION I: CLINICAL DEFINITION OF ALBINISM**
Chair: I.Gottlob, E.Piozzi

9.00-9.20  Expanding the phenotype of albinism?
B.Käsmann-Kellner

9.20-9.40  Phenotypic spectrum in FHONDA and albinism
M.Van Genderen

9.40-10.00  **Discussion**

10.00-11.30  **SCIENTIFIC SESSION II: CLINICAL AND MOLECULAR DIAGNOSIS OF ALBINISM**
Chair: K.Grønskov, S. Scalinci

10.00-10.20  Postnatal retinal development in albinism as compared to healthy controls
I.Gottlob
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<thead>
<tr>
<th>Time</th>
<th>Title</th>
<th>Speaker(s)</th>
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<tbody>
<tr>
<td>10.20-10.40</td>
<td>Visual cortex in albinism - anatomy and function</td>
<td>M.Hoffmann</td>
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<td>10.40-10.55</td>
<td>Contribution of electrofunctional examinations in albinism diagnosis</td>
<td>A.P.Tormene</td>
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<td>10.55-11.15</td>
<td>Molecular diagnostic approaches: Targeted sequencing, exome, array-CGH?</td>
<td>B.Arveiler</td>
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<td>11.15-11.30</td>
<td>Discussion</td>
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<td>11.30-12.00</td>
<td>Coffee break</td>
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<td>12.00-13.30</td>
<td><strong>SCIENTIFIC SESSION III: EUROPEAN DIAGNOSTIC EXPERIENCES</strong></td>
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<td>Chair: A.Del Longo, A.Taïeb, P.Primignani</td>
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<td>12.00-12.20</td>
<td>Clinical and molecular experience in Denmark</td>
<td>K.Grønskov</td>
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<tr>
<td>12.20-12.35</td>
<td>Clinical and molecular experience in Italy</td>
<td>L.Mauri, R.Terrana</td>
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<td>12.35-12.50</td>
<td>Albino Day in Madrid</td>
<td>M.Cortôn</td>
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<td>12.50-13.05</td>
<td>Albino Day in Bordeaux</td>
<td>F.Maurice-Picard</td>
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<td>13.05-13.30</td>
<td>Discussion</td>
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<td>13.30-14.40</td>
<td>Lunch break</td>
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14.40-16.00 **SCIENTIFIC SESSION IV: PATHOGENESIS OF ALBINISM**

Chair: L.Montoliu, M.V.Schiaffino

14.40-15.00 Understanding visual defects in albinism using mouse and cellular models
_A.Rebsam_

15.00-15.20 Syndromic albinism disorders affecting membrane trafficking during melanosome formation
_M.Marks_

15.20-15.40 Evidence for melanosomal channels and transporters critical for pigmentation
_E.Oancea_

15.40-16.00 **Discussion**

16.00-16.30 **Coffee/Tea break**

16.30-18.10 **SCIENTIFIC SESSION V: BEYOND EYES**

Chair: B. Arveiler, S.Penco

16.30-16.50 Oculocutaneous albinism in East and Southern Africa
_P.Lund_

16.50-17.10 Skin photoprotection issues in albinism
_A.Taïeb_

17.10-17.30 Dermatological characterization in Italian albinos
_D. Calò_

17.30-17.50 Audiological deficits in albinos: correlation with genetic/clinical phenotype
_G.Mingoia_

17.50-18.10 **Discussion**
8/4/2016

8.30-10.00  SCIENTIFIC SESSION VI
TECHNICAL SUPPORT AND THERAPEUTIC FRONTIERS

Chair: B.Käsmann-Kellner, A.Del Longo

8.30-8.45  Visual management training in low vision patients
F.Fabiani

8.45-9.05  Pharmacological therapy
B.Brooks

9.05-9.20  Defining safety and efficacy end-points for OCAI gene
therapy-based clinical studies
E.Surace

9.20-9.40  New animal models of albinism generated with CRISPR tools
L.Montoliu

9.40-10.00  Discussion

10.00-10.30  FINAL SCIENTIFIC DISCUSSION

Chair: I. Gottlob, A.Marocchi, L.Montoliu, M.V.Schiaffino

10.30-13.30  PLENARY FINAL SESSION

10.30-11.00  Summary of scientific presentations
B.Käsmann-Kellner , M.C.Patrosso

11.00-11.20  Summary of psycho pedagogical presentations
M. Nalli
11.20-11.50  Summary of association’s presentations  
   A. Glikshon

11.50-12.00  “Kindness for Kid”

12.00-12.10  Dermatological prevention  
   F. Brunetta

12.10-12.20  Photographic project “Every color of the White” presentation

12.20-13.30  Global discussion - Suggestions about guide lines on Albinism-  
  Proposal for the next meeting
ASSOCIATIONS SECTION

7/4/2016

8.00 - 8.30 Registration

8.30-9.00 Opening ceremony
Representative of the Region, G.P.Gesu, A.Marocchi, R.Masto, L.Paschetta, E.Tronconi

9.00-18.30 MORNING SESSION

9.00-10.00 Presentations of new online Associations

10.00-13.30 **Report Section on activities in different countries**
Defense of the Albino rights

11.45-12.10 Coffee break
Alliances with Public Administrations
Initiatives of social and scientific disclosure on this rare genetic condition
Albinism and technologies

13.30-14.40 Lunch break

14.40-18.15 EVENING SESSION
“Wina” Project Presentation: mobility and new frontiers

16.00-16.30 Coffee/Tea break
Albinism and European federation: state of art and new steps
Europe and Africa: alliances and projects
10.30-13.30  **PLENARY FINAL SESSION**

10.30-11.00  Summary of scientific presentations  
*B.Käsmann-Kellner, M.C.Patrosso*

11.00-11.20  Summary of psycho pedagogical presentations  
*M. Nalli*

11.20-11.50  Summary of association’s presentations  
*A. Glikshon*

11.50-12.00  “Kindness for Kid”

12.00-12.10  Dermatological Prevention

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

7/4/2016

8.00 - 8.30  Registration

8.30-9.00  Opening ceremony
Representative of the Region, G.P.Gesu, A.Marocchi, R.Masto, L.Paschetta, E.Tronconi

9.00-10.00  I SESSION
Chair: E. Tronconi

9.00-9.20  The special needs in the education of albino children
L.D’Alonzo

9.20-9.40  Verona response to the needs of young families: “Associative support desk for visually impaired children for family awareness and for the rehabilitation planning, after first diagnosis”
M.Nalli

9.40-10.00  Who benefited from the project Hypovision: Help Space.
Detailed quantitative and qualitative considerations
G.Zampieri

10.00-11.00  II SESSION
Chair: L. D’Alonzo

10.00-10.20  The experience at the Center for Pediatric Ophthalmology of Verona: the interaction between clinicians, families and educator of Help Space!
E.Gussone, I.Tomaello

10.20-10.40  Where the resistance to resilience are higher: support residential projects
E.Vlacos
10.40-11.00 From the analysis of the request to the understanding of child with visual impairment in the consultation stage at the Fondazione Robert Hollman
E. Mercuriali

11.00-11.50 III SESSION
Chair: M. Nalli

11.00-11.15 The multisensoriality as resource and educational strategy
M. Boneschi

11.15-11.35 A rehabilitative approach for integrating visually impaired child: the experience of the children low vision center IRCCS.E. Medea
G. Giammari, D. Valli

11.35-11.50 The experience of the Blind Institute of Milan: the educational activities for pupils with albinism in the context of services for visually impaired
F. Lisi

11.50-12.10 Coffee break

12.10-13.30 III SESSION
Chair: E. Piozzi

12.10-12.30 Dyslexia, pathophysiology and rehabilitation
S. Scalinci

12.30-12.50 The taking into care of the visually impaired child rehabilitation for different age groups: the intervention model of Chiossone Institute
E. Cocchi
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<tr>
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<tr>
<td>12.50-13.10</td>
<td>The achievement of the autonomy in the visual impaired person: can be done? S.Presti</td>
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<tr>
<td>13.10-13.30</td>
<td>The visually impaired student: strategies for school inclusion S.Benedan</td>
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<tr>
<td>13.30-14.30</td>
<td>Lunch break</td>
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</tbody>
</table>
| 14.30-18.30 | **ROUND TABLE**  
**STATE OF THE ART OF THE LAWS IN MATTERS OF INCLUSION**  
Map of the educational services for children with sensory disabilities offered by the region  
The usefulness of educational support Help Desk  
The services offered by CTS-NTD: the case of Como  
The alternation between school and work, an important tool for the life project of people with albinism  
The role and responsibilities of the supporting teacher  
An associative experience of welcome and service in Sicily  
School and social inclusion  
Discussion and Conclusion |
| 16.45-17.15 | Coffee/Tea break                                                     |
MEDICAL SECTION SPEAKERS

ARVEILER BENOÎT
Geneticist
Medical Genetic
University of Bordeaux, F

MARTA CORTÓN
Biomedical Research
Genetics and Genomics Department
Institute–Jimenez Diaz Foundation University Hospital, E

ALESSANDRA DEL LONGO
Ophtalmologist
Paediatric Ophthalmology
Niguarda Hospital, Milan, IT

FLAVIA FABIANI
Ophtalmologist
Hypovision Center
Ospedali Riuniti di Bergamo, IT

IRENE GOTTLOB
Ophtalmology
Ophtalmology Group
University of Leicester, UK

KAREN GRØNSKOV
Ophtalmogenetic Researcher
Cellular and Molecular Medicine
Kennedy Medical Genetics, DK

MICHAEL HOFFMANN
Ophtalmologist
Ophtalmology Department
University of Magdeburg, D
PATRICIA LUND
Geneticist
School of Life Science
Faculty of Health and Life Sciences, UK

BARBARA KÄSMANN-KELLNER
Ophthalmologist
Department of Ophthalmology
University of Saarland, D

MICHAEL S MARKS
Pathologist
Department of Pathology and Laboratory Medicine
Children’s Hospital of Philadelphia Research Institute, USA

ALESSANDRO MAROCCHI
Pathologist
“Emeritus Department Head”
Niguarda Hospital, Milan, IT

RODOLFO MASTO
Special Commissioner
Istituto dei Ciechi di Milano, IT

LUCIA MAURI
Geneticist
Medical Genetics
Niguarda Hospital, Milan, IT

GIUSEPPE MINGOIA
Otolaryngologist
ORL Clinic
Università di Milano Bicocca, IT

LLUIS MONTOLIU
Research Scientist
Department of Molecular and Cellular Biology
Centro Nacional de Biotecnologia, E
DONATA CALO’
Dermatologist
Medicine Department
Niguarda Hospital, Milan, IT

ELENA OANCEA
Researcher
Department of Molecular Pharmacology, Physiology and Biotechnology
Brown University, USA

LUCIANO PASCHETTA
Director
Istituto per la Ricerca la Formazione e la Riabilitazione, IT

MARIA CRISTINA PATROSSO
Geneticist
Ethics Committee
Niguarda Hospital, Milan, IT

SILVANA PENCO
Geneticist
Medical Genetic
Niguarda Hospital, Milan, IT

FANNY MAURICE PICARD
Paediatric Dermatologist
Department of Paediatric Dermatology
Bordeaux University Hospital, F

ELENA PIOZZI
Ophthalmologist
Paediatric Ophthalmology
Niguarda Hospital, Milan, IT

PAOLA PRIMIGNANI
Geneticist
Medical Genetic
Niguarda Hospital, Milan, IT
ALEXANDRA REBSAM
Researcher
Institut du Fer à Moulin
INSERM, F

SERGIO SCALINCI
Ophthalmologist
Low Vision Academy
Università degli Studi of Bologna, IT

MARIA VITTORIA SCHIAFFINO
Researcher
Group Leader, Dibit
San Raffaele Hospital, Milano, IT

ENRICO MARIA SURACE
Researcher
TIGEM
Telethon Institute of Genetics and Medicine

ALAIN TAÏEB
Dermatologist
Department of Dermatology
University of Bordeaux, F

ROBERTA TERRANA
Ophthalmologist
Paediatric Ophthalmology
Niguarda Hospital, Milan, IT

ALMA PATRIZIA TORMENE
President International Society for Clinical Electrophysiology of Vision, IT

ELISA TRONCONI
President ALBINIT, IT

MARIA VAN GENDERN
Ophthalmologist
Department of Visual Diagnostics
Bartiméus Institute for the Visually impaired, NL
PSYCHO PEDAGOGICAL SECTION SPEAKERS

SONIA BENEDAN
Tiflologa
Servizio socio-educativo
Centro Non Vedenti - Brescia

MARCO BONESCHI
Psicologo
Associazione Nazionale Subvedenti

FRANCO CASTRONOVO
Docente di Tecnologia
Area Sostegno alla Persona (DA, DSA, BES) - Como

ELENA COCCHI
Psicologa
Istituto David Chiossone per i Cechi e gli Ipovedenti
Centro di Riabilitazione delle Disabilità Visive

LUIGI D’ALONZO
Pedagogista
Centro studi e ricerche sul disagio e le povertà educative universitarie
Università Cattolica di Milano

ROCCO DI LORENZO
Presidente
Associazione Retinopatici ed Ipovedenti Siciliani - ARIS

rita garlaschelli
Insegnante
Ufficio scolastico per la Lombardia

giuseppina giammari
Neuropsichiatra Infantile
Centro Ipovisione dell’Età Evolutiva
IRCCS “E. MEDEA” – La Nostra Famiglia, (Lecco)
ELENA GUSSON
Oftalmologo
Istituto di Clinica Oculistica
Università di Verona

FRANCO LISI
Direttore Scientifico
Dirigenza Istituto dei Ciechi - Milano

GIOVANNI MERLO
Direttore
Lega per i Diritti delle Persone con Disabilità
Federazione Regionale Lombarda - FISH

MARCELLA NALLI
Pedagogista
Progetto vederesentire
SocioCulturale Cooperativa Sociale Onlus - Mestre, Venezia

SALVATORE NOCERA
Avvocato
Federazione Italiana per il Superamento dell’Handicap - Roma

STEFANO PAROLINI
Rappresentante
Settore Socio Educativo e Socio Assistenziale
SocioCulturale Cooperativa Sociale onlus - Mestre, Venezia

LUCIANO PASCHETTA
Psicologo
Federazione Associazioni Nazionali Disabili

LUCIA PELLEGRINI MARCHESINI
Rappresentante ALBINIT

ELENA PIOZZI
Oftalmologa
Oculistica Pediatrica
Ospedale Niguarda, Milano
SEBASTIANO PRESTI
Vice Presidente
Associazione Nazionale Istruttori Orientamento Mobilità Autonomia Personale
Brescia

SERGIO SCALINCI
Oftalmologo
Low Vision Academy
Università degli Studi of Bologna

ELENA MERCURIALI
Psicologa
Fondazione R. Hollman - Padova

ILARIA TOMAELLO
Ortottista
Oculistica
A.O. Universitaria Integrata di Verona

ELISA TRONCONI
Presidente ALBINIT

DANIELA VALLI
Pedagogista
Associazione La Nostra Famiglia
IRCCS “E.Medea”, (Lecco)

ELENA VLACOS
Psicoterapeuta
Servizio di Neuropsichiatria Infantile
Fondazione Istituto Neurologico Nazionale “C. Mondino”, (Pavia)

GIOVANNI ZAMPIERI
Pedagogista
Ce.Di.Sma
Università Cattolica di Milano
REGISTRATION

Medical section
Physicians, Biologists and Researchers (only for operators in the albinism sector)

ECM credits: 8.40

Online Registration: albinit.org/progetti/IIIEDA

Maximum number of participants: 80

Registration cost:
• 250,00 € for Physician, Biologist or Researcher (information regarding included services on (http://albinit.org/progetti/IIIEDA/)  
• 80,00 € for Student – Fellow - Laboratory technicians  
• free registration for internal employees

The meeting will be held in English

Psyco pedagogical section
Orthoptists, Speech therapists, Laboratory technicians, social and health educators

ECM credits: 4.90 (credits will be awarded to teachers and University students)

Online Registration: albinit.org/progetti/IIIEDA

Maximum number of participants: 200

Registration cost:
• 80,00€  
• free registration for internal employees

L’incontro si terrà in italiano.
SCIENTIFIC ADVISOR

ELENA PIOZZI
Paediatric Ophthalmology Director
Niguarda Hospital, Milan, IT

SECRETARIAT

Maria Cristina Patrosso
cristina.patrosso@gmail.com

HOW TO REACH THE MEETING

Lombardy Region Building can be reached with MM2 (Gioia stop)