



Ospedale Niguarda

Sistema Socio Sanitario



Regione  
Lombardia

ALBINIT 



OSPEDALE SAN RAFFAELE

APRIL, 7<sup>th</sup> AND 8<sup>th</sup>, 2016

# III EDA 3th 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 (IILEDADA).*

*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  
**Psycho 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

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**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*

- 10.20-10.40** Visual cortex in albinism - anatomy and function  
*M.Hoffmann*
- 10.40-10.55** Contribution of Electrofunctional Examinations in Albinism  
Diagnosis  
*A.P.Tormene*
- 10.55-11.15** Molecular diagnostic approaches: Targeted sequencing, exome,  
array-CGH?  
*B.Arveiler*
- 11.15-11.30** *Discussion*
- 11.30-12.00** *Coffee break*
- 12.00-13.30** **SCIENTIFIC SESSION III:  
EUROPEAN DIAGNOSTIC EXPERIENCES**
- Chair: A.Del Longo, A.Taïeb, P.Primignani
- 12.00-12.20** Clinical and molecular experience in Denmark  
*K.Grønskov*
- 12.20-12.35** Clinical and molecular experience in Italy  
*L.Mauri , R.Terrana*
- 12.35-12.50** Albino Day in Madrid  
*M.Cortón*
- 12.50-13.05** Albino Day in Bordeaux  
*F.Maurice-Picard*
- 13.05-13.30** *Discussion*
- 13.30-14.40** *Lunch break*

**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  
*M.C.Negri*

**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 OCA1 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  
*F. Glickson*
- 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

# ASSOCIATIONS SECTION

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**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



**8/4/2016**

**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  
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the general public-Proposal for the next meeting

# PSYCO 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  
*V.Schoch*
- 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*

- 12.50-13.10** The achievement of the autonomy in the visual impaired person: Can be done?  
*S.Presti*
- 13.10-13.30** The visually impaired student: strategies for school inclusion  
*S.Benedan*
- 13.30-14.30** Lunch break
- 14.30-18.30** **ROUND TABLE**  
**STATE OF THE ART OF THE LAWS IN MATTERS OF INCLUSION**
- Chair: F.Castronovo, R.Di Lorenzo, R.Garlaschelli, G.Merlo, S. Nocera, S.Parolini, L.Paschetta, L.Pellegrini Marchesini
- 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 CORTÒN**

Biomedical Research  
Genetics and Genomics Department  
Institute-Jimenez Diaz Foundation University Hospital, E

## **ALESSANDRA DEL LONGO**

Ophthalmologist  
Paediatric Ophthalmology  
Niguarda Hospital, Milan, IT

## **FLAVIA FABIANI**

Ophthalmologist  
Hypovision Center  
Ospedali Riuniti di Bergamo, IT

## **IRENE GOTTLOB**

Ophthalmology  
Ophthalmology Group  
University of Leicester, UK

## **KAREN GRØNSKOV**

Ophthalmogenetic Researcher  
Cellular and Molecular Medicine  
Kennedy Medical Genetics, DK

## **MICHAEL HOFFMANN**

Ophthalmologist  
Ophthalmology 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 Biotecnología, E

**MARCO ANTONIO NEGRI**

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

**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



# PSYCO 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- Genova

## **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  
Grande Ospedale Metropolitano 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

### **VITTORINA SCHOCH**

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

Fondazione CIRNA ONLUS

Centro Studi Terapia della Gestalt - Milano

### **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](http://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](http://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  
*crisrina.patrosso@gmail.com*

## HOW TO REACH THE MEETING

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

## Patronages



Milano

## Supported by



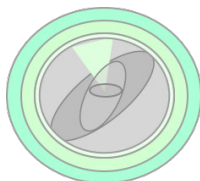
Associazione Italiana Ortottisti  
Assistenti in oftalmologia  
[www.aiorao.it](http://www.aiorao.it)

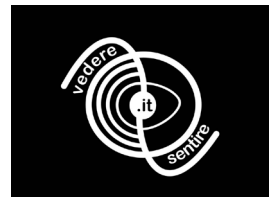
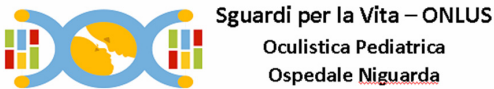


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# CeDisMa

Centro studi e ricerche  
sulla Disabilità e Marginalità







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