

il giornalino

delle famiglie

ONLUS



Associazione
Nazionale
di Volontariato
Cornelia
de Lange June 2005



Fondazione
Pierfranco e Luisa Mariani
neurologia infantile ONLUS

English version



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The Italian Association has its Scientific Committee



Responsible: Scientific Coordinator
Dr. Angelo Selicorni

Eight years after its foundation and very close to the first World Congress held in Italy, the Italian Association has made another important step forward: the creation of its Scientific Committee.

What is a Scientific Committee?

It is an consultation and support organ which has the double function of to make proposals and to express opinions on all potential projects regarding the study, research, assistance to people with CDLS which in some form involves the association.

It further operates as an interface to the scientific and academic world for the Association.

What does a Scientific Committee do?

The members of the Scientific Committee meet periodically (at least once or twice per year) to discuss about the best and most concrete measures to further the research and to improve the assistance of the CDLS patients in Italy.

It will be the responsibility of the members of the Scientific Committee to give CDLS a priority position among their scientific interests, be it in terms of clinical or research activity be it in terms of continuous education.

These people will further be available, each one in his/her area of expertise, to respond to or to take direct responsibility for specific questions families or professional might have.

An additional important role of the members of the Scientific Committee will be to develop and to deploy, always in direct agreement with the Associations board, all educational as well as informational projects with scientific scope which the Association believes to be important to be planned and realized; they will be available for active participation.

Whom is our Scientific Committee composed of?



dr Emanuele Basile, psycho-pedagogic and family therapist, IRCCS E. Medea Association "La Nostra Famiglia" from Bosisio Parini (Milan province), is a member of the International Federation's SAC (Scientific Advisory Council) and

has been supporting the association for many years. His areas of expertise regard the educational approaches for people with CDLS and the emotional-psychological challenges for the families.

dr Sergio Luzzani,

pediatric surgeon and endoscopist; always connected with our Association and recent member of the International Federation's SAC. His area of big expertise refers to all internal surgical issues in relation to the syndrome, in particular with reference to the diagnosis and the treatment of the gastro-esophageal reflux.



prof.ssa Paola Marchisio,

pediatrician at the I Pediatric Clinic of the University of Milan.

Prof. Marchisio has also recently joined the SAC of the International Federation. As pediatrician she focuses on the follow-up of people with CDLS with specific competence on the aspects of the upper respiratory apparatus.



dr Luigi Memo,

clinical genetic and neonatology specialist, recently joined the SAC of the International Federation.

As genetic specialist his particular areas of expertise are the diagnosis of the syndrome and the characteristics associated with the syndrome in the follow-up of patients.

As neonatology specialist he will contribute his specific knowledge to all problems and complications of the first weeks/months of life which often represent difficulties and reasons for deep preoccupation of the parents of a baby with CDLS.

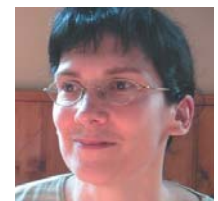


dr Gioacchino Scarano,

medical genetic expert and pediatrician. Aside from his diagnostic capacity and his expertise with treatment protocols, Dr. Scarano will contribute with his specific

competence on the subjects of genetic consultancy and prenatal diagnosis of CDLS, in particular in view of the recent promising discoveries in relation to the underlying genetic defect.

dr.ssa Laura Villa, children neuropsychiatrist, IRCCS E. Medea Association "La Nostra Famiglia" from Bosisio Parini (Milan province). Thanks to the collaboration between the Association and the institute where Dr. Villa works, she



is certainly one of the Italian children neuro-psychiatrists who has had the opportunity to evaluate the largest amount of people with CDLS. Consequently her competence lays in the areas of rehabilitation and training programs with particular interest and competence in behavioral aspects.

And *last but not least*: **Dr. Angelo Selicorni**, medical doctor, specialized in Pediatrics and Medical Genetics. He works in Milan in the "I Clinica Pediatrica dell'Università di Milano in the pediatric department. He is the chief of a group of medical geneticists whose work is to diagnose at clinical level, children with various genetic syndromes. Angelo is deeply involved in the follow-up of these children with particular attention towards some syndromes: CdLS is the best of these. He is involved in some projects in clinical research to better understand the natural history of various conditions.

The first official meeting of the Scientific Committee will take place on Wednesday, June 15th, 2005, in occasion of the World Conference. A second, more operative and decision oriented meeting is planned to take place in Fall of this, together with the Association's board meeting.

I would like to take the opportunity to thank all the colleagues who have given their availability to accept this additional role and who certainly will do their best to contribute to the development of our Association and to improve the assistance of the people with CDLS in Italy.

What is the Federation CDLS?

The Italian association is part of the Federation of national CDLS organisations united by Cornelia de Lange Syndrome. Other member countries are: Australia, Canada, Denmark, France, Ireland, Japan, Italy, Netherlands, Poland, Portugal, United Kingdom and the United States. The Federation has been founded in 2001 and counts approx. 2.500 members.

The members of the Federation have a shared mission to offer help, give hope, and reach out to persons with CDLS and those who love and care for them.

The main functions of the Federation are:

- 1) to liaise with and "manage" the International Scientific Advisory Council - SAC (in the context of their insistence that they work for us),
- 2) to be a forum of best practices in serving our CDLS families (exchanges ideas and information and organize an international conference every two years)
- 3) to act as a resource base for the wider family of people affected by CDLS throughout the world for both research and for families moving from one country to another.

For additional information see:
www.cdlsworld.com



What is the Scientific Advisory Council (SAC)?

The Scientific Advisory Council is made up of medical and other professionals who take responsibility for the shared knowledge of specialties. Its goal is to help families, researchers and other professionals understand the complexities of the syndrome.

The Council also includes "Professional Directors" from countries around the world who represent the Federation among their own professional communities and are a focal point for SAC information within that country.

SAC Chairman:

Alex Levin (Canada)

Emeritus Professor:

Laird Jackson (USA)

Specialties:

Education

Psychology

Behaviour

Psychiatry

Speech & Language/feeding

Ophthalmology

Research Genetics

Clinical Genetics

ENT/Audiology

PT/OT Orthopaedics

Neurophysiologist/Neurology

Radiologist

Dentistry

Structure and Program

Wednesday 15th June

03:00 pm Italian Scientific Council Meeting
05:00 pm International Scientific Council Meeting

Thursday 16th June

02:30 pm International Federation Council Meeting
Thursday 16th June and Friday 17th (until noon)

Professional Day

Friday (starting with lunch)

to Sunday 19th June

International Family Congress

Thursday 16th June 2005

Professional day

8,30 a.m. – 9 a.m.
Welcome and introduction

8,15 a.m.
Start of Childcare

9 a.m.–10,45 a.m. General Session
The diagnosis of Cornelia de Lange Syndrome (CDLS)

History of the syndrome

Raul Hennekam, Olanda

Clinical aspects and phenotype variability

Angelo Selicorni, Italia

History of the gene search

Laird Jackson, Usa

State of the art in the research of the genetic defect

Emma Tonkin, Gran Bretagna

NIPBL mutation spectrum in CDLS

Matthew Deardorff, Usa

10,45 a.m.–11 a.m. **Coffee break**

11 a.m.–12,30 p.m. General Session
Medical problems of people affected by CDLS

Gastro-esophageal reflux and CDLS

Sergio Luzzani, Italia

Eye problems

Alex Levin, Canada

Upper airways infections and hearing

Paola Marchisio, Italia

Orthopedic problems

Antonio Memeo, Italia

Seizures and EEG findings in Cornelia de Lange syndrome

Tiziana Granata, Andrea Van Lierde, Italia

12,30 p.m.
(or end of session)
End of childcare + pick up children

1 p.m. **Lunch**

1,50 p.m.
Opening registration desk
Start Childcare

2 p.m.– 3 p.m. General Session
Psyco-motor and cognitive development of the CDLS child

Psyco-motor development of CDLS child: a rehabilitative approach
Laura Villa, Italia

Communication development in the CDLS: how to stimulate for speech and language skills

Marjorie Goodban, Usa

3 p.m. – 4 p.m. General Session

Educational and behavioural aspects of CDLS

The behavioural phenotype of CDLS

Chris Oliver, Gran Bretagna

Education and the student with CDLS

Mary Morse, Usa

6 p.m.

4 p.m. – 4,30 p.m. **Coffee break**

Closure registration desk

4,30 p.m.

Brief communication on clinical researches and assistance experiences

6,30 p.m.

(or end of session)

End of childcare + pick up children

6,30 p.m. - 8 p.m.

6,30 p.m. **Winding up**

"Punto anamnesi"

8 p.m. **Dinner**

Friday 17th June 2005

8,30 a.m. – 10,30 a.m.

Workshops for specific professionals

8,15 a.m.

Start of Childcare

*Opening registration desk**

Educational approach

Emanuele Basile, Italia, Mary Morse, Usa

Speech and language stimulation

Cheri Carrico, Usa, Marjorie Goodban, Usa

Clinical follow-up

Alex Levin, Canada, Sergio Luzzani, Angelo Selicorni, Italia

Behavioural problems

Chris Oliver, Gran Bretagna, Laura Villa, Italia

* The registration and information desk will remain open the whole day

10,30 a.m. – 11 a.m. **Coffee break**

11 a.m. – 12,30 p.m.

Brief communication on clinical researches and assistance experiences

12,30 p.m. **Winding up**

Alex Levin, Canada

12,30 p.m.

(or end of session)

End of childcare + pick up children

1 p.m. **Lunch**

Families Congress

Friday 17th June 2005 afternoon

2 p.m. **Welcome and introduction**

Moderator Family Congress:

Michela Vuga

2,30 p.m.–4,15 p.m. General Session

News and updates:

2 p.m.

Start of Childcare

CDLS family and genetic journeys

4 p.m. - 6,30 p.m.
"Dishappyland"
corner

(The registration
and information
desk will remain
open the whole day)

6,30 p.m.
(or end of session)
End of childcare +
pick up children

Closure registration
desk

6,30 p.m. - 8 p.m.
"Punto anamnesi"

8,30 p.m. - 9,30 p.m.
"Dishappyland"
corner

Laird Jackson, Usa
**Clinical severity and prognostic
index**

Angelo Selicorni, Italia
Medical complications
Alex Levin, Canada

4,15 p.m. - 4,45 p.m. **Coffee Break**

4,45 p.m.-6,30 p.m. General Session
From theory to practice 1:

**Stimulation of CDLS child in the first
year of life**

Laura Villa, Italia
**Procedures for developing speech
and language in CDLS**

Marjorie Goodban, Usa
**Feeding issues in children with
Cornelia de Lange syndrome**
Cheri Carrico, Usa

7,45 p.m. **Dinner**

9,30 p.m. "International Ice Cream
Cabaret & Karaoke"

Saturday 18th June 2005

7 a.m. - 8.30 a.m. **Breakfast**

9 a.m. - 11 a.m. General Session
From theory to practice 2:

8 a.m. - 9 a.m.
"Punto anamnesi"

8,30 a.m.
Start of Childcare
Opening registration
desk

**Behavioural problems in children and
adults with CDLS.**

**A psychological perspective on cau-
ses and treatment**

Chris Oliver, Gran Bretagna
**Ideas, ideas and more ideas for tea-
ching students who have CDLS**
Mary Morse, Usa

Everyday life of adults with CDLS
Francesca Atzeri, Giovanna Oliosio,
Anna Veronese, Italia

11 a.m. - 11,30 a.m. **Coffee Break**

11,30 a.m. - 1 p.m. General Session
**Assisted autonomy - experience and
projects**

Vacation project Italy
Lorenza Breda, Tiziana Zarpellon,
Italia

Life in small community
Alan Peaford, Gran Bretagna
**Post-school programs -
the Australian example**
Jenny Rollo, Australia

12,30 p.m.
(or end of session)
End of childcare +
pick up children

Closure registration
desk

1 p.m. **Lunch**

2,30 p.m. - 4,45 p.m.
Roundtable
**Children with CDLS and school expe-
rience: experiences in different
countries**

Moderator Roundtable Discussion:
Susanna Del Gamba, Italia
4,45 p.m. - 5,15 p.m. **Coffee break**

5,15 p.m. - 6,30 p.m.
**Concurrent workshops for parents
and siblings**

**The relationship between parents of
a child with CDLS**

Emanuele Basile, Italia
**Affected children and healthy
siblings: parents challenge**
John Morse, Usa

**Siblings workshop - for siblings aged
5 and over**

Bernadette Dalingwater, Canada
**Adults with CDLS - planning for their
future**
Julie Mairano, Antonie Kline, USA

7,45 p.m. **Dinner**

9,30 p.m. "Gala Sweet Gala"

Sunday 19th June 2005

7 p.m. - 8.30 p.m. **Breakfast**

9 p.m. - 9,30 p.m. General Session
**The International programm of
motor activity and sport "directed"
to the intellectual disability: Special
Olympics**

Alessandro Palazzotti, Italia

9,30 p.m.-10,30 p.m. General Session
Non conventional therapies

Musicotherapy and CDLS

Antonia Madella Noja,
Lorenza Vigorelli, Italia
Horsetherapy
Danièle Raymonde Nicolas Citterio,
Italia

10,30 p.m. Closing of Congress
**The future of the Scientific Advisory
Council**

Advisory Council
**The present and the future of the
Federation Council**
(Newly elected Federation Chairperson)

**Preview of the next International
CDLS Conference**
Bernadette Dalingwater, Canada

12 a.m. **Winding up**
Giorgio Amadori

1 p.m. **Lunch**

3,30 p.m. **Optional sight seeing trip
to Massa Marittima**
(for who has booked in advance)

2 p.m.

Start of Childcare
2,30 p.m.
Start of individual
consultation

Opening registration
desk

4 p.m. - 6,30 p.m.
"Dishappyland"
corner

2,30 p.m. -6,30 p.m.
"Punto anamnesi"

6,30 p.m.
(or end of session)
End of childcare +
pick up children

Closure registration
desk

7,45 a.m.

Church Service
8 a.m. - 12,30 p.m.

"Punto anamnesi"
8,30 a.m.

Start of Childcare
Opening registration
desk

10 a.m. - 12,30 p.m.
"Dishappyland"
corner

12,30 p.m.
(or end of session)
End of childcare +
pick up children

Closure registration
desk

2,30 p.m. - 6,30 p.m.
Individual
consultation

"Punto anamnesi"
"Dishappyland"
corner

“ P u n t o A N A M N E S I ”

Check-in for Individual Consultations

In order to use the time during the individual medical consultations as efficient as possible, a check-in point called “**Punto ANAMNESI**” has been setup.

Each expert requires some general information about the patient before being able to properly and effectively respond to the parents’ questions. In order to best utilize the 20 minutes of the single consultation, it is opportune that this information be collected beforehand. Therefore **a parent should come to the check in point as soon as possible, however by all means before the first appointment**, where a doctor will collect all important and required information on a respective form in English.

This sheet will be given to the parents who obviously must take it to each single doctor’s appointment. By reading the form, the expert will be able to obtain in a few seconds a complete overview of the patient and will consequently be better able to respond to the parents’ questions.

At the end of the meeting the specialist is asked to summarize the information provided to the family in the lower part of the form.

The purpose is twofold: give to the parents a minimum written feedback of the meeting and inform the following colleagues of what has already been discussed.

After the last doctor’s appointment the parents should return to the check-in point where they will receive a copy of the form.

This entire work will help to collect all information during this conference and in the future the same document can be used to update it at the next national or international conferences.

The check-in point “**Punto ANAMNESI**” is located in the lower floor (**UNDERGROUND**), on the right side of the registration and information desk, in the area where the individual consultations will take place.

Operating Hours:

Thursday	6:30pm – 8:00pm
Friday	6:30pm – 8:00pm
Saturday	8:00am – 9:00am; 2:30pm – 6:30pm
Sunday	8:00am – 12:30am; 2:30pm – 6:30pm

INDIVIDUAL DOCTORS’ CONSULTATIONS

The individual doctors’ meetings take place in the **UNDERGROUND** floor, on the right side of the information and registration desk.

The “Check-in” point “Punto ANAMNESI” will indicate the proper area.

Appointment Hours:

Saturday 2:30pm – 7:30pm
Sunday 9:00am – 12:30pm; 2:30pm – 6:30pm

As every year, each family will have the opportunity to have individual consultations with specialists of various areas.

The meetings – **which are strictly free of charge** - will last a maximum of 20 minutes each. In case the expert does not speak the language of the requesting family, a translator (at least Italian/English) will be provided with the aim to optimize the results of the meeting.

It is always recommended to have precise questions to be asked to each specialist ready, hence obtaining better results from the consultation.

Starting Thursday afternoon it will be possible to check the complete calendar of all consultations on

a big chart close to the information and registration area and the check-in point., where, in case necessary, changes will be published.

IMPORTANT

In case there are still openings, you have the possibility to sign up for additional consultations on the chart.

We also would like to inform you that we have the possibility to offer consultations with the dentist, Dr. Doug Clemens, for many years member of the SAC.

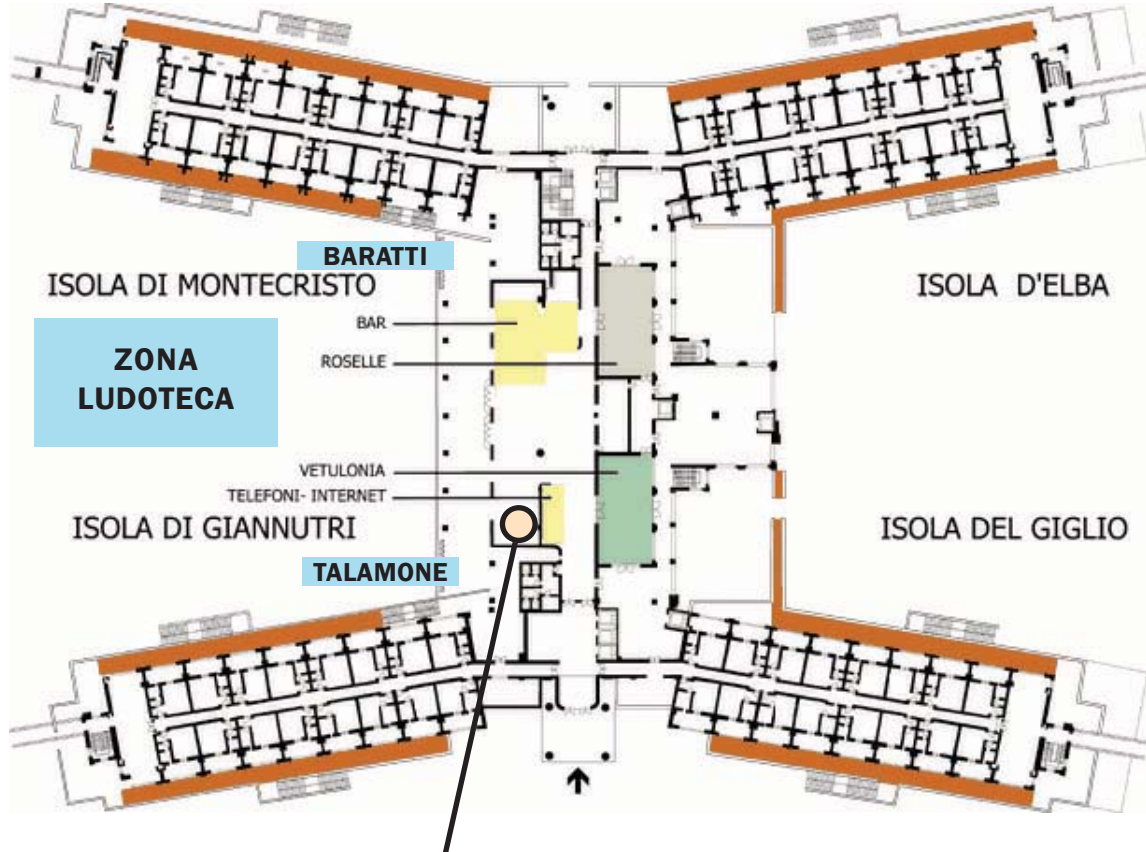
Please pay attention not to overwrite or accidentally move or remove consultation already scheduled with the new ones.



Fattoria “La Principina” Center Congress



GROUND FLOOR



“ DISHAPPYLAND ” * Information Corner

* A project financed with funds from the “Osservatorio Nazionale del Volontariato” - Social and Labor Ministry.

In the conference folder you will also find a CD entirely dedicated to the project “DISHAPPYLAND” (DISabled peoples’ HAPPY LAND). We have also organized an information kiosk, where you can obtain any information required and where you can get a feeling for the “spirit” which has animated all participants of this vacation, by seeing and touching small big objects realized by our kids. A PC will continuously show photos and film clips from this beautiful vacation.

The “DISHAPPYLAND” kiosk is located on the **GROUND FLOOR** in the general registration area of the hotel, behind the phone booths.

The kiosk is manned:

Friday: 4:00pm – 6:30pm; 8:30pm – 9:30pm
Saturday: 4:00pm – 6:30pm
Sunday: 10:00am – 12:30pm; 2:30 – 3:30pm

CHURCH SERVICE

*If you wish to attend
a catholic church service, a mess
is planned to be held*

**on Sunday morning,
at 7:45am.**

Signs will indicate the location.

CHILDCARE / "LUDOTECA"

When we first came to visit the beautiful "Fattoria la Principina" a few months ago,

the entire Pesaro Village staff got excited about transforming this place into a giant playground, full of games, initiatives and activities with fun times as well as relaxing moments for the duration of the conference.

Thanks to the Italian volunteers and the American Girl Scouts, starting 8:30 am every morning there will be no "truce" ... for children and adults that are ready to get involved in our great events.

Some activities will be available all day (e.g. swimming pool, inflatable games), while others (e.g. make-up for children, "amusement park") will be organized as special events and activities.

Everything starts in "Sala BARATTI" at 8:30 am and goes on – with a planned break for lunch – until the end of the conference sessions, right before dinner.

What about evening activities and events? Don't miss the "International Ice Cream Cabaret & Karaoke" on Friday night or the "Gala Sweet Gala" on Saturday evening!

SCHEDULE

Thursday, Friday, Saturday, Sunday
From 8:30 AM to 12:30PM
From 2:00PM to 6:30 PM
(childcare service is assured until the end of the session)



ACTIVITIES

9,30 AM - 12,00 AM Friday Saturday Sunday	SWIMMING POOL ACTIVITIES INFLATABLE AREA GAMES
15,00 PM - 18,00 PM Friday Saturday	CHILDREN TRACTORS CIRCUIT INFLATABLE AREA GAMES
16,30 PM - 18,00 PM Friday	"AMUSEMENT PARK" (Games with final prize) "AREARTE" (creative lab)
16,30 PM - 18,00 PM Saturday	MAKE UP CORNER with background for photos and immediate printing
sempre aperta Friday Saturday Sunday	Cinema or music room

Other games or sport activities will be organized and scheduled directly by the staff.

We remind all families to pick up their children at the end of the conference sessions, please be punctual.

CHILDCARE - "PASS"

All activities will take place at the **GROUND LEVEL**.
 The respective area is in front of the entrance hall of the hotel.

BARATTI and TALAMONE rooms:

One of these rooms which is accessible from the inside with a direct passage to the outside will be the base for the "drop-off" of the children to the childcare staff.

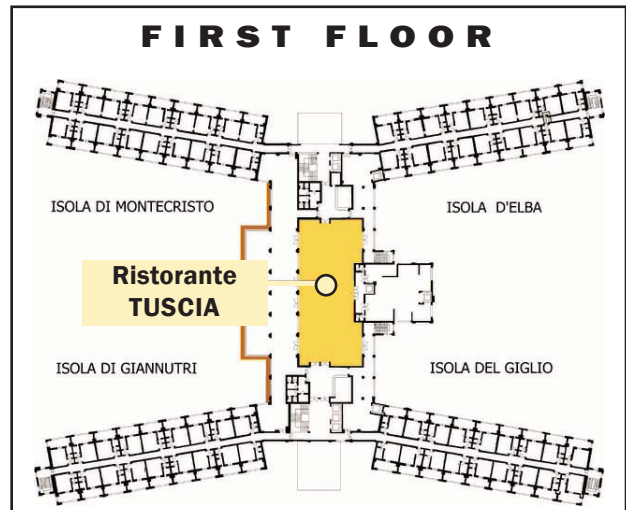
When you drop you child off you will receive a **PASS** with the name of the child and his/her identification number (which will be the same as you have on your badge).

Keep the PASS safely, as it will be the identification you need to pick up your child at the end of the session or at any time you deem necessary.

If necessary you may give the PASS to a person of your trust to have your child picked up.

Remember: your child will be given to the person who has the PASS of the child.

On the **FIRST FLOOR**
is the "Tuscia" restaurant located
where the speakers and the organizers
of the Mariani Foundation eat.



On the **SECOND FLOOR**
is the "Ertruria" restaurant located
where all participants of the congress,
the families and volunteers eat.

For the conference participants staying at the hotel the cost of the meal is included in the room rate, meal coupons which must be presented in order to access the restaurant will be found in the conference folder.

MEAL COUPONS

Who does **STAY OVERNIGHT** at La Principina, but joins the conference, meal coupons to access the "Ertruria" restaurant **may be purchased at the registration and information desk of the Association** (20 Euro each).

*We hope that you will find
this experience fulfilling.*

*Please do not hesitate to ask
for assistance
at our information desk*

Enjoy it!

Angelo, Antonella, Simona, Ulla



We thank the Grosseto Province for its kind sponsorship and support and in particular for providing the bus for transporting the CdLS-Families inside "Fattoria La Principina" premisses.

With the kind support:

- Ministero del Lavoro e delle Politiche Sociali*
- Regione Toscana*
- Comune di Grosseto*
- Società Italiana di Pediatria*
- Società Italiana di Genetica Umana*
- Società Italiana di Neonatologia*
- Gruppo di Studio di Genetica Clinica della Società Italiana di Pediatria*
- Gruppo di Studio di Genetica Clinica della Società Italiana di Neonatologia*
- International Center for Birth Defects, Roma*
- Istituto Superiore di Sanità*

- I Clinica Pediatrica dell'Università di Milano*
- U.O. Neurologia dello Sviluppo*
- Istituto Nazionale Neurologico Carlo Besta*