

## NEWSLETTER DECEMBER 2005

Dear partners , friends and relatives, also the year 2005 is going to finish and we can affirm that this year, too we worked hard!

We promised to use this way of communication more frequently and punctually but unfortunately our human resources, all voluntary service, did not allow us to maintain promises; in any case we reached the aim to wish you in time a Merry Christmas and Happy 2006.

By this newsletter we shall review together what achieved in 2005 and shall get an eye in next 2006.

On the achievement of any target we say “ stop now for a short rest....” but the will to do is so great and things to do so important and urgent that we never maintain that intention!

We cannot never forget, however, that the achievement of important targets have been possible thanks to two fundamental elements, indispensable also for the future, that is to say :

- The necessary economic resources always obtained thanks to the generosity of the several persons, companies and institutions who help us

**and, firstly,**

- The indispensable collaboration and the confidence of the numerous families all over the world who help and support us in spite of the heavy engagements in following a disabled son. Without their help all what achieved would not have been possible!

Just for thanking you, parents, we report, herebelow, a letter from a mother of a child suffering IDIC15 syndrome who, in a few words, described clearly and simply our life.

“

### WHERE ARE THE PARENTS?

They are on the phone with doctors and hospitals, they are fighting with insurance companies against bureaucracy for trying to answer their child's necessities.

They are submerged by papers and accounts for medicines, they are trying to find a sense for a system which seems to be made for confusing and frightening with the only exception of the more experienced ones.

They are at home to change the napkins to their son 15 years old or to try to lift from the w.c. their daughter of 37 Kgs, they are spending one hour a meal to feed a child who cannot masticate, they are changing medications, catheters, oxygen distributors.

They are patiently seated and waiting in hospital rooms while their child try to recover from an operation or a heavy epileptic crisis.

They are sleeping in turn since their child does not sleep more than two or three consecutive hours by night.

They stay closed at home since often family and friends, may be too shy or missing will, don't help them by staying sometimes with their child while institutions, which should help those parents, are not in a position to help them owing to internal problems due to financing cuts.

Where are the parents?

They are trying to spend some time also with their children, fortunately not disabled; they are fighting with their partner for saving a marriage, because misfortune not always joins together; they are doing two and even three jobs for covering the expenses which are higher and higher. Sometimes one only parents is doing all that.

Where are the parents?

They are trying to survive in a society speaking a lot but doing nothing; they are trying to recompose their broken dreams for reaching, at least, a normal way of leaving for their children and family. They have so much to do..... trying to survive!

“

In such a way, step by step, herebelow what we have realized for our children:

## SCIENTIFIC PROJECTS

### PROJECT OF GENETIC/MOLECULAR RESEARCH

The first research doctorate financed by our association on last April 2004, was addressed to the genetic/molecular field at the Catholic University of Rome under the organization and coordination of (lady) Prof. Marcella Zollino and Prof. Giovanni Neri, both members of our Scientific Committee.

Our researcher, (lady) Dr. Laura Seminara, <http://space.comune.re.it/assring/Ricercatori.htm> is collecting the blood samples of the children suffering all the genetic aberrations connected to chromosome 14 (ring, delations, translations, duplications,...) as well as the blood samples of their parents to analyse them through sophisticated methodologies (FISH) and to define, better than by the traditional cariotype, quantity and seat of the genetic material missing in each single patient and, eventually, in his parents.

Such a reasearch has the specific target of identifying the genes involved in the different aberrations of chromosome 14 verifying their eventual responsibility about its presence or not and the higher or lower gravity of the signs and symptoms of the clinical picture.

Much to our satisfaction, we invite you to read the first scientific documents drawn up by our Scientific Committee reported in our web site obtained thanks to the researches finaced by us,

<http://space.comune.re.it/assring/Pubblicazioni.htm>

Those documents (abstract and poster) were presented in 2005 in two important medical congresses: The National Congress of Italian Society of Human Genetic S.I.G.U. and the American Congress of American Society of Human Genetic A.S.H.G.

We warmly thank all families, not only in Italy but all over the world, who enthusiastically participated in this research and we invite all the families who not yet did it to contact us for sending the blood samples necessary to that important study.

You can find further details on that project and how to participate in our site:

<http://space.comune.re.it/assring/Ricerca.htm>

### DATABASE PROJECT

We already sent from September a questionnaire to all families (an other one will follow for a deeper search on the base of the answers given in the first one) to be filled up, even with the help of their family doctor, to be returned to us completed with pictures and medical reports.

We know it is a very long and compelling work, sometimes even painful because it obliges to relive difficult moments faced by our sons and our family, but we beg you, dear parents, just for our sons, to collaborate even in this project having the following target: to collect a complete and homogeneous selection of data on patients suffering aberrations of chromosome 14 in order to allow our searchers to obtain the more of information allowing a correct definition of correlation between genotype and fenotype.

This very important instrument has been personalized for our patients by (lady) Dr. Angela Scarano, children's Neuropsychiatrist, working at the Children's Neuropsychiatry Complex Structure of "Santa Maria Nuova" Hospital of Reggio Emilia whose Director, Dr. Della Giustina, is member of our Scientific Commitee. (Lady) Dr. Scarano is enjoying since February 2004 a three-year scholarship.

<Http://space.comune.re.it/assring/Ricercatori.htm>

Our database, XML form, runs by a personalized software bought from Ylichron Srl ([www.ylichron.it](http://www.ylichron.it)) and will allow, through a web access protected by password, to enter in real time into a number of tables containing all medical data concerning the patient (considered from all mediactal points of view), offering a complete control of the clinical case. A searching motor will allow to cross and to correlate the data of more patients contained in the Database.

What above in order to improve medical information on those syndromes for a better use of medical, surgical and rehabilitating therapies for our sons.

In accordance with Italian law we have also enclosed a form of authorization to deal medical data which you are kindly requested to return duly signed with the questionnaires.

## PROJECT OF CLINICAL STUDY OF CHILDREN SUFFERING CHROMOSOME PATHY 14

This project avail itself of a CLINICAL AND INSTRUMENTAL PROTOCOL which will be developed at the Children's Neuropsychiatry Complex Structure of "Santa Maria Nuova Hospital" – Reggio Emilia (Director Dr. Elvio Della Giustina) and at the Children's Neuropsychiatry Complex Structure of "Ospedale Maggiore" in Bologna (Director Dr. Giuseppe Gobbi); both Directors are members of our Scientific Committee.

**DIAGNOSTIC APPROACH AND TARGETS:** Problems of children suffering chromosomepathy 14 concern specific genetic aspects, neurological-neuropsychological and behaviour development, epilepsy even precocious and medicine resistant and chronic rehabilitating necessities. The target is to obtain that the "condition" actually rare and unknown, can be from now on better studied and known

That protocol will foresee, therefore, the hospitalization of the children for a few days in order to submit them to instrumental and diagnostic tests. That project, too has the aim to help the doctors to better understand all the symptoms connected to those rare syndromes and to help and to take better care of our "special" children.

That protocol will start on January 2006; our Association will be in a position to welcome one or two family per month. We well know the difficulties you will face for participating in that project, the fears, the doubts you will have in travelling with your children, but this is a very very important opportunity we offer you for trying to better understand these rare genetic syndromes.

Therefore, we invite all interested families to get in touch with us for confirming their availability to participate and the more convenient month; afterwards the families will be put in communication with (lady) Dr. Scarano for verifying all tests already effected by the child and to complete the medical investigation.

As regards the many foreign families collaborating with us, we are working for solving in the shortest possible delay the bureaucratic problems concerning the repayment of effected tests, but in the meantime we accept their adhesion.

We have drawn up a detailed document explaining all tests which will be made and their purpose as well as the operative and logistic details of the stay (which will be at the charge of the association).

That project has been possible thanks to our financing of a scholarship to (lady) Dr. Scaranto and to a research doctorate which will be assigned on next March 2006 to a Researcher Doctor who will work at the Children's Neuropsychiatry Complex Structure of "Ospedale Maggiore" of Bologna – Director Dr. Giuseppe Gobbi – member of our Scientific Committee.

<http://space.comune.re.it/assring/Ricercatori.htm>

## FAMILIES WITH CHILDREN SUFFERING "RING 14" AND "DELATION 14" SYNDROMES

Step by step we are getting more and more.

**Now we are 127 families** of which:

- 60 with "RING 14"
- 57 with delation chromosome 14 or different aberrations

Our site has been up-dated with the insertion of a lot of new children, who is in possession of the password can enter in the reserved area called "OUR CHILDREN'S HISTORY"

<http://space.comune.re.it/assring/Ring14.htm>

You will find the histories and the addresses. Who is not in possession of the password but wish to visit our site can ask it by mail to our Association.

## UP-DATE WEB SITE

Further to what previously informed, the main novelty is that in the chapter MEDICAL AND SCIENTIFIC RESEARCH we have inserted three new subjects:

CONGRESSES:

<http://space.comune.re.it/assring/Congressi.htm>

where you will find detail of the congresses organized by our association with relevant documents

SCIENTIFIC PUBLICATIONS

<http://space.comune.re.it/assring/Pubblicazioni.htm>

where you will find all scientific articles, abstracts and posters published by our researcher doctors

OUR RESEARCHER DOCTORS

<http://space.comune.re.it/assring/Ricercatori.htm>

where you will find the presentation and mail addresses of our researcher doctors.

## FUND COLLECTION

## **PINNACOLO TOURNEY**

On November 17, 2005 the Lions Club Canossa Val d'Enza organised a Pinnacolo Tourney at the Tennis Club Albinea (Reggio Emilia) on favour of "Ring 14" Association.

In spite of the various similar initiatives which take place in our town a lot of people participated in the initiative which obtained a good result. Many thanks to the people attending to the initiative and to the organisers of the same. A special thank to Marisa, wife of the President of the Club, Dr. Prampolini, who took care of the whole organization with great passion and delicacy. It is already the third year that the Lions Clubs give us their support which, further to the important economic aid, makes us very proud of the great demonstration of esteem for our work.

## **CHRISTMAS CARDS**

As for last years we printed the Christmas cards which have been sold both to private persons and to companies. A special thank to the employees of Marella company who participated generously in the initiative and to the following companies : "Santa Maria Nuova" Hospital, "Nuova Planetario S.p.A.", "Strolin-Strhold" Group, Sintex.

## **SOLIDARITY STALLS**

Also this year, braving the cold and inclement weather (it was even snowing!) we punctually were present at two appointments in Reggio Emilia : on November 24<sup>th</sup> - Patron holidays and also on Saturday December 10<sup>th</sup>. It was the handsomer stalls of last years, we collected more than thousand Euro!

We wish to thank heartily :

- All the volunteer ladies who relaized the wonderful objects for sale working night and day for us
- All the volunteers who braved the intense cold (particularly Monica) from 7 a.m. to 8 p.m. for selling our objects
- All very willingly people who bought our object

and, uncredible, there are persons who made all what above (Monica and Stefania)! A great solidarity chain!

## **NEXT STALLS**

They will take place at the Children School "P. FREIRE" of Canali on the occasion of Christmas holidays respectively on December 15<sup>th</sup> - 20<sup>th</sup> and 21<sup>st</sup>.

We wish to thank the Direction of Reggio Emilia Schools and Children Schools for the opportunity given to us.

Collected funds will be destined to purchase furnishings for the Children Neuropsychiatry Department in Reggio Emilia in the context of the project "ADORN THE NEUROPSYCHIATRY", in collaboration with the Schools and municipal Children Schools of Reggio Emilia, "Reggio Children". Coopselios and Friends of Reggio Children. We wish to thank them heartily for the availability shown. That project will be described in detail in next newsletter.

## **WHO WANTS TO SUPPORT OUR PROJECTS FROM AN ECONOMIC POINT OF VIEW:**

can do it by a free transfer to the following bank coordinates:

**UNICREDIT BANCA SPA**  
**COUNTRY IT - CIN EUR 27 - CIN H - ABI 02008 - CAB 12820**  
**C/C NR. 3665310 - in favour of "ASSOCIAZIONE INTERNAZIONALE RING 14"**

In the next newsletter which, we promise, will be publicated within March 2006, we will illustrate the future projects and we will thank all people who supported us, from an economic point of view, during the year.

**We take the opportunity for wishing you a Merry Christmas and a Happy New Year !**