



# MISSION REPORT

2012/13 Balance - 2014/15 Budget

  
**ring14**

**ITALIA**

ENGAGEMENT, SUPPORT, FUTURE  
**ONLUS**



## RING14 ITALY

Was founded as a non-profit organization in Reggio Emilia in May 2002 by a group of families with children afflicted by the syndrome of chromosome 14, a rare and currently incurable genetic disorder.

**The main symptoms are overwhelming: severe drug-resistant epilepsy, severe language delay, malformations of the oro-gastric apparatus, repeated and severe respiratory infections, significant psychomotor retardation, behavioral disorders and autistic-like microcephaly.**

In centers located in Philadelphia, Boston, Strasbourg, Milan, Pavia and Genoa, Ring14 Italy is currently sponsoring research and clinical projects on genetics, the study of language and autistic traits, which are greatly assisted through the use of our biobanks and databases. We also organize international workshops for physicians and researchers, support families with information services and assistance, as well as create opportunities for developing friendships and sharing experiences.

*I am at your complete disposal for any further information you desire about our activities and how you can become a part of our organization.*

*Stefania Azzali*

The mother of Matteo  
Vice-President of Ring14 Italy

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Note

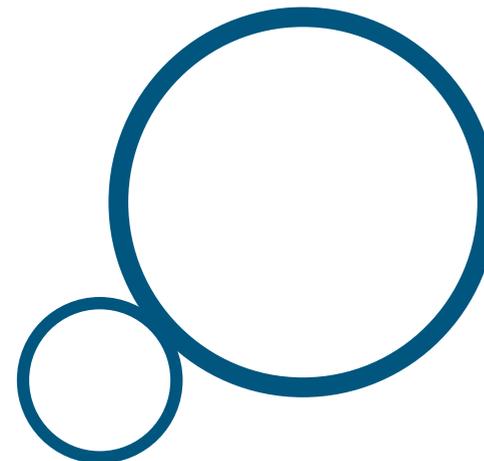
# WHY A BUDGET

A budget is necessary in order to document **if the activities** undertaken by Ring14 Italy **conform to organizational aims prescribed in the statutes**, as well as offer readers a straightforward, but comprehensive instrument to evaluate the work of our association.

The Mission Report takes into account both the **balance for the biennium 2012/2013** and the **budget for 2014/2015**. This report covers a time span much longer than a normal accounting period for non-profit organization in order to provide a comprehensive view of the average working and planning period of Ring14 Italy.

The document is divided into five sections, each of which explores different aspects of our association:

- About us
- 2013 Results
- Our Projects
- Our resources
- Our goals for the future



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Our **Solidary Biscuit** project is a joint initiative of Ring14 Italy in collaboration with the restaurant Cà Matilde di Reggio Emilia. The proceeds will finance research regarding epilepsy.



*About us*

# RING 14 ITALY

## BOARD OF DIRECTORS

Andrea Russo | President  
Stefania Azzali | Vice-president and Treasurer  
Francesca Pivetti | Secretary  
Massimiliano Braglia | Member  
Corrado Fiscale | Member  
Francesco Giglioli | Member  
Lorenza Mazzi | Member  
Gabriella Prodan | Member  
Antonella Rizzi | Member

**Marie,**  
afflicted with the syndrome  
of chromosome 14,  
suffers from seizures and her  
drug-therapy has to be changed  
continuously.

One of the research projects of  
Ring14 is aimed at studying  
stem cells in order to identify  
the most effective  
pharmacological  
strategy.



## SCIENTIFIC COMMITTEE

Prof. Giovanni Neri  
Prof. Emilio Perucca  
Dr. Giuseppe Gobbi  
Prof. Marcella Zollino  
Dr. Elvio Della Giustina

# RING14 MILESTONES ONE STEP AT A TIME

Ring14, founded in Reggio Emilia, becomes a volunteer non-profit Association and our **Scientific Committee** is established.

2002

The first **PhD** in Genetics at the Catholic University of Rome is financed and Ring14 becomes part of **Eurordis, the European network on rare diseases**.

2004

The first project studying **language** delay conducted by the University of Milan Bicocca is financed.

2007

The second **International Congress** is organized, on this occasion the first results of our scientific studies are presented.

2008

• First association in Italy to reach an agreement with the **Telethon Genetic Biobank Network** for the collection and storage of biological samples for scientific research.

• The **American Journal of Medical Genetics** publishes our first scientific article and it devotes its cover to Ring14.

• **Ring14 France** is opened.

2009

• **The first International Workshop** involving the best researchers from around the world is organized.

• The fairy tale "**The child from the ring**", a metaphor of disability and social integration is published.

• The newspaper **Sole 24 Ore** publishes an article about us.

• **Ring14 USA Outreach** is founded.

2011

• **Telethon finances** a project on the syndrome of chromosome 14 at the University of Pavia.

• The first **Charity Dinner** with **chef Bruno Barbieri**, attended by many supporters is held.

• **Epilepsia** publishes our scientific research project.

• **More than 140 articles about us**, including one of **Vanity Fair**, are published in local and national press.

2013



2014

**RING14 International**, a coordination network between the various local offices is founded.

• **www.ring14.org** is created to enhance communication **among the hundreds of families from 10 countries**.

• **Fund raising activities** to finance scientific research are begun.

2003

• **Collection of clinical data** of our children is organized into a database.

• **A project for psychological assistance** to families is financed.

2005

• **The first International Congress** is organized involving families from all over the world and mobilizing hundreds of volunteers. It is the first time that the scientific community addresses the disease.

• **A study on epilepsy** in collaboration with the hospital Maggiore in Bologna is financed.

2006

• We participate for the first time the **Telethon**.

• **Epileptic Disorder** publishes our study on epilepsy.

2010

• We celebrate our **first ten years**.

• The first research projects in **peer review**: a Philadelphia (Spinner) e a Strasburgo (Herauld) are financed.

• The **Children Hospital di Boston** studies pro-bono our Syndrome.

• **Clinical L&P** publishes our project.

2012

## 2015/2017 WITH YOUR HELP OUR NEXT STEPS

• Open **new chapters abroad** to improve assistance to families

• Start **studies on autistic traits** to identify the best rehabilitation therapies

• Fund new **scholarships** and **international research grants**

• Carry out initiatives set out by the **International Workshop** to involve the best researchers in the world

## OUR FAMILIES

Ring14 is the only reference point for **hundreds of families around the world**: we are present on 5 continents, in 23 countries and we communicate in 5 languages.

Our main **goal** is to **help families overcome their isolation**, encourage them in times of fear, give them useful medical information concerning the care of their child and try to give them hope, without creating false expectations. This disease is very serious and dangerous: the more families are informed and aware, the more they can help their child.

**We are always there for them**, be it online, on the phone or in person during those fun events we organize every year.

We continuously receive new contacts via our website or social network, we respond immediately, and we offer all our support. Sometimes families call us after just having received the diagnosis, still shaken by the tremendous change that they will soon have to face. Other times, they contact us years later, when the children have grown and the pain and initial shock has diminished somewhat.

Getting in touch with the association and with other families can be a big help, but it also requires a great emotionally effort and courage. You can start to see what the future will be like for you, for your family and your child. You can feel and share the pain of others. You can be consoled. It can be said that **the association is a large give-and-take container of emotions**. We must be ready to handle them all, for good or bad.



# RING14 VOLUNTEERS AND FRIENDS

Many people help and assist us by dedicating their time and expertise in support of our many activities.

Some help us **by managing fundraising initiatives**, such as fair stands, dinners, shows; others volunteer a few hours a week at our office and **perform operational and secretarial tasks**.

We also have professionals who provide their specific expertise. Just think about the large group of volunteers who translate our literature and news briefs in four languages. There are cooks and waiters who help us organize charity dinners. Actors, comedians and singers who organize new and interesting events. We count on an array of computer experts, photographers, accountants, lawyers, who through their pro-bono work allow us to save resources that can be allocated better to financing our projects.

Among these we would like to specially thank:

- the law firm **Hogan Lovells**, Milan;
- the **Studio Dalla Salda** which handles our payroll;
- **Studio Ligabue** which does our translating;
- the actor **Claudio Santamaria**;
- the chef **Bruno Barbieri**;
- the artist **Enrico Magnani**;
- the conductor and journalist **Daniele Piombi** who helped us produce the advertisement to publicize tax deductible donations.



*In the big picture in a clockwise direction: the Parish of San Marco (Reggio Emilia) for our Charity Dinner; staff of volunteers for solidarity stands; Monica, Marisa, Mary, Lorenza and Francesca.*



*Researchers invited to attend the  
2nd International Scientific Workshop organized by Ring14 Italy - 25/27 October 2014*



# THE FIGURES

Ring14 Italy balance sheet is an annual report finishing on December 31 of each year. For any further information, you can contact our office by writing to [info@ring14.it](mailto:info@ring14.it)

## CASH FLOW MANAGEMENT

<i>Revenues</i>	<i>31 dec 2013</i>	<i>31 dec 2012</i>
1) From ordinary activities		
1.1) Membership fees	€ 1,225	€ 1,550
1.2) Tax deductible contributions	€ 26,177	€ 26,000
Total income from ordinary operations	€ 27,402	€ 27,550
2) Financial and equity		
2.1) Income from bank deposits	€ 2	€ 1
Total financial income and equity	€ 2	€ 1
3) From fundraising		
3.1) Private donations	€ 54,267	€ 61,822
3.2) Companies donations	€ 36,475	€ 34,965
3.3) Contributions from organizations and foundations	€ 21,272	€ 39,005
3.4) Stands and local initiatives	€ 57,758	€ 25,319
Total income from fundraising	€ 169,770	€ 161,110
4) Other income		
4.1) Other income	€ 3	€ -
4.2) Extraordinary income	€ 1,924	€ -
Total income from ordinary operations	€ 1,927	€ -
<b>Total income</b>	€ <b>199,102</b>	€ <b>188,661</b>

<i>Expenses</i>	<i>31 dec 2013</i>	<i>31 dec 2012</i>
1) From ordinary activities		
1.1) Payments to employees and consultants	€ 42,699	€ 23,763
1.2) Fundraising expenses	€ 62,064	€ 51,586
1.3) Overheads	€ 8,287	€ 20,499
1.4) Depreciation	€ 1,887	€ 1,732
1.5) Other operating expenses	€ 321	€ 1,766
Total expenses from ordinary operations	€ 115,256	€ 99,345
2) Financial and equity		
2.1) Bank expenses	€ 405	€ 502
Total financial expenses	€ 405	€ 502
3) On projects		
3.1) Charges on projects	€ 43,341	€ 88,911
Total costs of projects	€ 43,341	€ 88,911
4) Income taxes		
4.1) IRAP (regional tax on productive activities)	€ 913	€ -
Total taxes	€ 913	€ -
<b>Total expenses</b>	€ <b>159,915</b>	€ <b>188,758</b>
<b>Positive operating result</b>		
	€ <b>39,187</b>	€ -
<b>Negative operating result</b>		
	€ -	€ <b>97</b>
<b>Total to draw</b>	€ <b>199,102</b>	€ <b>188,661</b>

The **total income for 2013** amounted to **199,102 euros**, thanks largely to income from ordinary operations (membership fees, tax-deductible contributions) and fundraising activities that constitute the largest share at 169,770 Euro. The **total expenses** were **159,915 euros**, which covered the costs of running projects and fundraising activities and paying employees. We close with a **positive operating result** of **39,187 euros** that will be used in 2015 to finance projects.

# HOW ARE RESOURCES USED

## 2013 EXPENSES (FOR ALLOCATION OF FUNDS)

■ Overheads	€	10,874	5%
■ Pocket expenses for Fund Raising	€	51,941	26%
■ Personnel expenses for Fund Raising	€	17,000	9%
■ Awareness initiatives	€	10,123	5%
■ Projects and scientific consultation	€	69,977	35%
■ Earnings from future projects	€	39,187	20%
<b>Total</b>	€	<b>199,102</b>	<b>100%</b>

**Zoe**  
is a little girl who meets the world every day with a smile and enthusiasm. She needs help so that her epilepsy doesn't ruin her life.



## RESEARCH AND SUPPORT FINANCED THROUGH YOUR HELP

Ring14 Italy is funding **scientific projects** on a national and international level, in clinical, genetic and drug therapies, to understand the pathogenesis and identify potential treatments for syndromes of chromosome 14.

**Based on the Telethon model**, all projects are evaluated and selected using the “**peer review**” system or by an international commission of scientists in order to avoid any possible conflict of interest.

The Association also organizes **international workshops** involving researchers on the highest level, experts in the field of cytogenetics, molecular genetics, clinical applications and, in particular, epilepsy with the aim of generating ideas and innovative approaches to the study of the disease.

In addition to supporting scientific research, we use our resources to establish

**social activities** aimed at ensuring support, help, and assistance in coping with the consequences of the disease.

The Ring14 Italy approach has always had an essential foundation. We consider **the family as a “place” where changes**, involving and benefiting all members can be fostered.

Below is a summary of the management of project funds for 2012/2013:

### PROJECT REPORT

### 2012/2013

Genetic Research (Prof. Spinner and Herauld)	€ 73,869	€ 35,287
Language development	€ 11,625	€ 7,577
Biobank	€ 7,513	€ 8,112
Workshop	€ -	€ 2,813
Vacations	€ -	€ 16,188
<b>Total</b>	€ <b>93,007</b>	€ <b>69,977*</b>

\* Compared to the previous year, the 2013 total is to be considered net of the Ring14 USA contribution, which, in 2013, paid directly research projects the amount of € 30,000.



**650.000 €**

is the total amount of  
**resources collected**  
by Ring14 in its  
10 years of activity

# RESEARCH PROJECTS IN DETAIL

## 1 • ANALYSIS OF GENE EXPRESSION IN SYNDROMES OF CHROMOSOME 14

Head: **Prof. Nancy Spinner** - Children's Hospital of Philadelphia at the Perelman School of Medicine at the University of Pennsylvania USA

**Duration:** 2 years (2013-2014)

**Cost:** € 130,000 - Funded by Ring14 Italy & Ring14 USA

The laboratory of Dr. Nancy Spinner at Children's Hospital of Philadelphia has been studying chromosomal rings since 2006 and is working to verify the hypothesis that the ring structure alters the expression of genes. Comparison between normal cell lines and those containing rings will help reveal their differences and explain how these relate to epileptic seizures. The hope is that discovery of the underlying seizure mechanism associated with ring chromosome can lead to a cure.

## 2 • CREATION OF A LABORATORY MODEL WITH CHROMOSOME 14 SYNDROME TO BETTER UNDERSTAND THE HUMAN SYNDROME

Head **Prof. Yann Herault** - Institut Clinique de la Souris (ICS) in Strasbourg

**Duration:** 2 years (2013-2014)

**Cost:** € 100,000 - Funded by Ring14 Italy, Ring14 USA and Institut Clinique de la Souris (ICS) in Strasbourg

The research objective is to build an experimental model to be used in the laboratory to study the altered molecular mechanisms in the syndrome linked to chromosome 14 and come to understand why and how they trigger symptoms such as epilepsy and behavioral and cognitive disorders.



## 3 • IPS STUDY ON CHROMOSOME 14

Head: **Dr. Anne Cherry** - Harvard University

**Duration:** 1 year (2013) - **Cost:** pro-bono work, equivalent to about € 50,000

IPS cells (induced pluripotent stem) are human cells that are manipulated to return to an undifferentiated stage. Such cells can, after subsequent manipulation, acquire specific characteristics and become, for the experimental purposes, cells of the liver, eye, heart...

Thanks to this project, IPS cells were obtained from chromosome 14 syndrome patients. In the future, these cells may be used for therapeutic trials.

## 4 • CHROMOSOME 14 SYNDROME: TOWARDS A DETAILED CORRELATION BETWEEN GENOTYPE AND PHENOTYPE

Head: **Prof. Orsetta Zuffardi** - University of Pavia

**Duration:** 2 years (2014-2015) **Cost:** € 118,000 - **Funded by TELETHON**

The project starts from the assumption that epilepsy, the most serious and intractable phenomenon in patients with syndromes of chromosome 14, is due to the loss of end parts of the chromosome. Through a study of molecular genetics, patients with abnormalities of chromosome 14 of different entities will be compared. Hopefully, the region of chromosome 14 whose absence causes the outbreak of the disease will be identified.

In a second phase of the project, starting with IPS cells, or neuronal cells, neurons will be obtained on which to test new drugs in the laboratory.

## 5 • BIOBANK AND CLINICAL DATA BASE

Head: **Dr. Chiara Baldo** - Galliera Genetic Biobank TGBN

**Duration:** since 2009 to present - **Cost:** € 8,000 per year - Funded by Ring14 Italy

Biobanks are authorized centers to preserve genetic material of patients and make it available to researchers around the world. In December 2009, Ring14 Italy stipulated an agreement with the **Telethon Genetic Biobank Network** for the collection of blood and skin samples of their patients and their families. This Biobank contains more than 300 biological samples available to the international scientific community.

Ring14 also runs a database of clinical data are collected from children which is useful to understand which symptoms are related to this syndrome, in order to stimulate and develop transactional research.

## 6 • LANGUAGE DEVELOPMENT IN CHILDREN WITH CHROMOSOME 14 ABERRATIONS

Head: **Dr. Laura Zampini** - Department of Psychology of the University of Milan / Bicocca

**Duration:** 5 years (2010-2014) - **Cost:** € 75,000 - Funded by Ring14 Italy

The project aims to describe the linguistic abilities of children with this syndrome and has already led to some interesting results showing that children lacking a greater amount of genetic material do not necessarily have a more compromise development.

Instead, those with autistic traits have a less favorable outcome for language and psychomotor development. These results emphasize the importance of early detection of the presence of autistic traits in order to strengthen as much as possible communication skills.

# INTERNATIONAL SCIENTIFIC PUBLICATIONS

2014 - **Clinical Linguistics & Phonetics**

*"Developing with ring 14 syndrome: a survey in different countries"*

2013 - **Epilepsia**

*"Epilepsy in ring 14 syndrome: a clinical and EEG study of 22 patients"*

2012 - **Clinical Linguistics & Phonetics**

*"Linguistic and psychomotor development in children with chromosome 14 deletions"*

2012 - **European Journal of Medical Genetics**

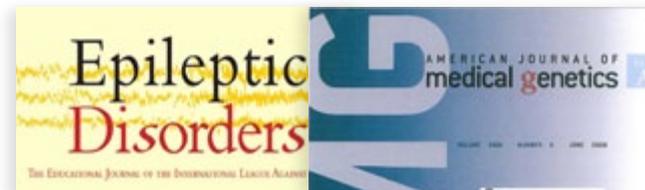
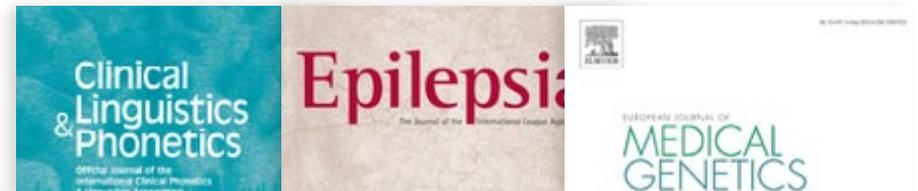
*"The ring 14 syndrome"*

2010 - **Epileptic Disorder**

*"Partial epilepsy complicated by convulsive and nonconvulsive episodes of status epilepticus in a patient with ring chromosome 14 syndrome"*

2009 - **American Journal of Medical Genetics**

*"The ring 14 syndrome: clinical and molecular definition"*



To review the articles visit our website [www.ring14.org](http://www.ring14.org)

## SERVICE AND SUPPORT TO FAMILIES

### PSYCHOLOGICAL COUNSELLING AND FAMILY SERVICES

Since the beginning of our activity, we have carried out a service of *Psychological Assistance and Family Counselling*. The service is dedicated to children and their families in order to assist them during the challenging period of communication of the diagnosis and in dealing with the day-to-day difficulties of the disease.

### “THE STORIES OF OUR CHILDREN” PROJECT

A narrative medicine project, *“The Stories of our Children”* are a collection of children’s stories written by their parents, so that our individual experiences can be shared with all of us.

### INTERNATIONAL SCIENTIFIC CONVENTION

For years, we have promoted opportunities for meetings organized for families around the world on the occasion of our *International Scientific Convention*. This enables our families to meet each other, to feel accepted and not “different” or “unusual”. It is an excellent opportunity to exchange information, emotions, difficulties, losses, successes, to share moments of joy and serenity, but also have a chance to listen and learn from the (few) medical experts on our syndrome.

### HOLIDAY PROJECT

In 2009, we started to offer families an experience of a relaxing holiday. Holiday locations were chosen specifically to meet not only the “special” needs Ring 14 children, for example the presence of a medical staff ready to respond to any need or emergency, but also to include their healthy siblings. In this way, all the family can enjoy fun activities together.

### SIBLINGS PROJECT

An opportunity to dedicate attention, companionship and interaction to children and teenagers who have severely disabled brothers or sisters does not happen frequently. This is the idea behind this project.

The project includes many activities such as dinners and trips together, group discussions and meetings, with the coordination and support of educators and psychologists. In 2009, this experience culminated in a week aboard a Navy ship, an exceptional environment for strengthening group bonding.

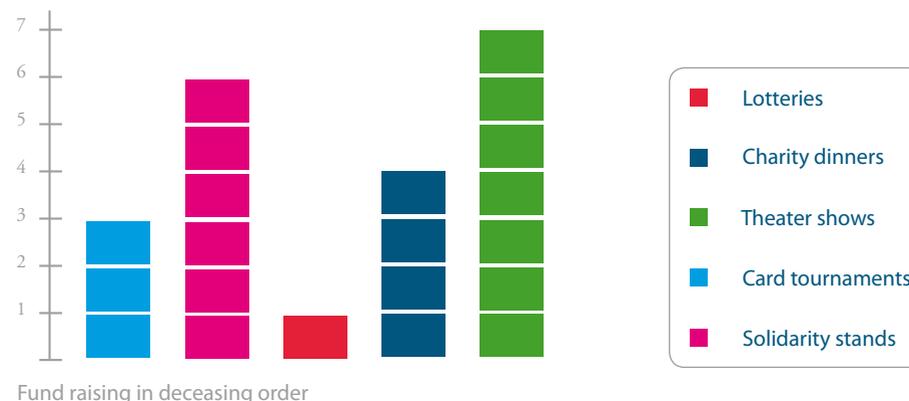


# WHO SUPPORTS US?

## REVENUE 2013 (BY SOURCE OF FUNDS)

Private	€	139,427	71%
Company	€	36,475	18%
Public Bodies	€	2,500	1%
Grant foundations	€	12,702	6%
Non-profit	€	6,070	3%
Other revenues	€	1,928	1%
<b>Total</b>	€	<b>199,102</b>	<b>100%</b>

## FUND RAISING INITIATIVES 2013



Numerous companies have helped us during the year through donations of goods or economic support.

In particular, we would like to point out the important initiative taken by the company **Rewind**. The company allowed their employees to make a monthly donation in our favor in the form of payroll giving. Their donations were then matched by the company. € 25,000, equivalent to half the annual cost of a grant for the financing of a scientific research project, was collected.

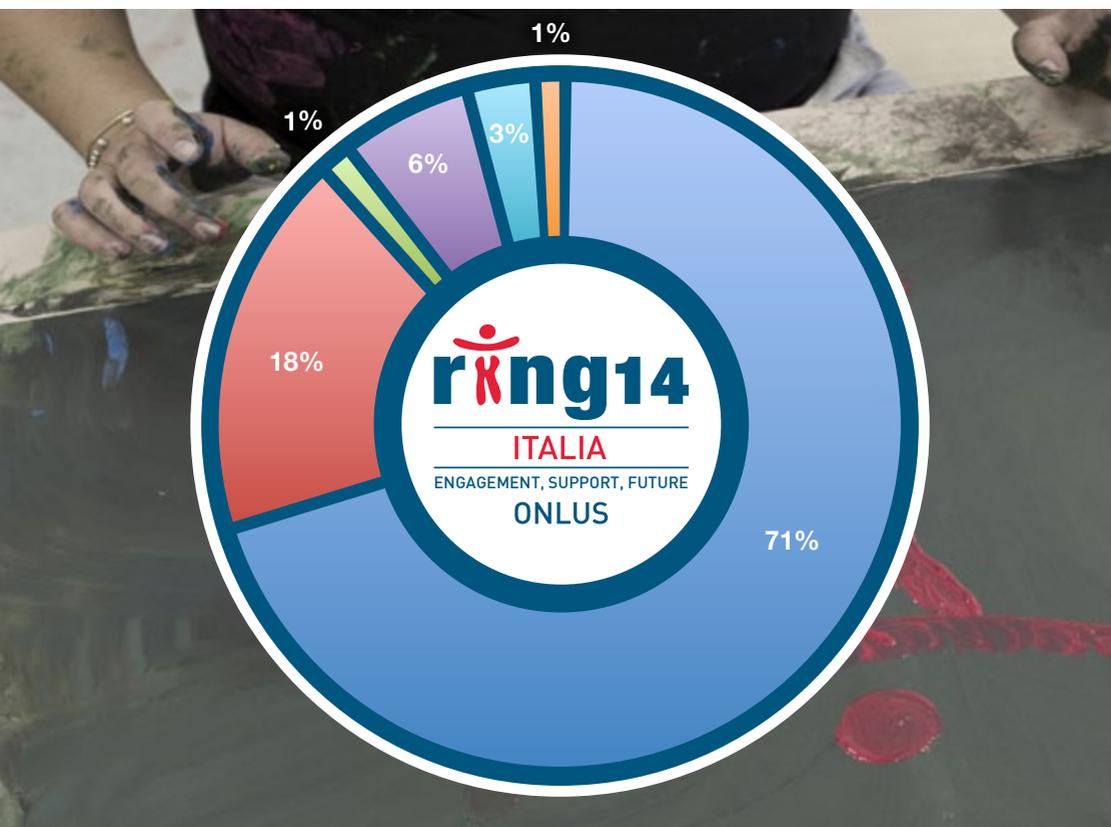
The company **Easy-call** has decided to allocate a percentage of each call made from their call center to our association.

The Michelin-star restaurant **Cà Matilde** di Reggio Emilia, thanks to the participation of chef Andrea Incerti Vezzani and his wife Marcella, have created and produced Ring14 solidary biscuits, which they offer to their customers and supply to various Ring14 activities. All proceeds from sales go to Ring14 Italy. **Pellicceria Garavaldi** gave us a percentage of sales;

**Fondazione Pietro Manodori** has consistently supported our scientific projects since 2003;

**Fondazione UniCredit**, through the involvement of an extensive network of employees, has sustained us for several years through GiftMatch.

Many companies support us by donating their products to our initiatives. These include: **Pluricart, Casali Vini, Lusvardi Wine, Salfrutta, Righi, Smeg, Diffusione Tessile, Melville.**



## FORECASTED EXPENSES AND COSTS

### FORECASTED PROJECTS COSTS 2014/2015

Language project	€ 8,000	€ 8,000
Study project on autism	€ -	€ 50,000
Biobank project	€ 11,500	€ 10,000
Holidays project	€ 5,500	€ 5,000
Workshop project	€ 23,000	€ -
Scientific grant project	€ 50,000	€ 75,000
Data base project	€ 3,000	€ 2,000
Overseas locations development project	€ -	€ 10,000
<b>Total</b>	<b>€ 101,000</b>	<b>€ 160,000</b>

As shown in the table, for the current year **an increase in expenses** for projects has been forecasted (from € 69,977 to € 101,000 - see report p. 21).

For 2015, the projected increase is **58%** compared to 2014. The increased resources will cover the following:

- activation of a new research project in collaboration with the University of Trento, relative **to the study of autistic traits of syndrome of chromosome 14**;
- **opening of new offices abroad**, to improve on-site assistance to families;
- refinancing of research projects in progress that have obtained best overall results.



#### Sunny

came from France to participate in a Ring14 holiday.

For our families, it is important to meet and learn from each other. We have a lot of fun together too.





## *how to help us*

*We can achieve all our projects thanks to the support of people who believe in us.*

### HOW TO MAKE A DONATION:

- by bank or money transfer

UNICREDIT - IBAN: IT 27 H 02008 12820 000003665310

SWIFT CODE: UNCRITM1OR0

- by credit card

Go to our website: [www.ring14.it/eng](http://www.ring14.it/eng)  
and click on "Donate Now"

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