

RING14 HELP & RESEARCH FOR CHILDREN AFFECTED BY RARE GENETIC DISEASES – ONLUS

A step forward in the genetic research

Within 2014 Professor Nancy Spinner is engaged in an in-depth study on the missing genes in patients affected by Ring 14.

This is the second international grant financed by RING 14 Italy, RING 14 USA and Diffusione Tessile.

A new international study for RING 14

The International Organization RING14 Onlus is relentless. Every day it works in order to achieve its goals : make the scientific research become a tool that looks at the future granting a better life to the children affected by chromosome 14 syndromes.

For RING14, 2012 has been an important year due to the different paths the international studies have started with, to which we have to add a new project that will be achieved in 2014, representing the **second international grant**, financed by RING14 Italy, also thanks to the support of RING14 USA.

We are talking of the genetic study "[Gene expression analysis in the Ring 14 syndrome](#)", performed by **Professor Nancy Spinner, Director of the Philadelphia Children's Hospital in partnership with Professor Giovanni Neri, Director of the Istituto di Genetica Medica of Università Cattolica of Rome**. This study will last **2 years** and will be supported by innovative technologies available only in a few centres worldwide. Its main goal will be to study in depth the function of the missing genes in patients affected by Ring14, in order to understand where the symptoms like epilepsy and psychomotor retardation come from and to identify the genetic medicaments able to cure those genes.

Understand the causes of epilepsy and detect rational treatments

Professor Spinner's research is based on the hypothesis that **the ring-shaped chromosomes in children affected by Ring 14 might be the cause of the clinical symptoms**. In fact the ring-shaped chromosomes are associated to an overlapping of phenotypes that include growth retardation, mental disability and serious epileptic seizures, difficult to control even in the absence of deletions or Ring 14 associated duplications. Professor Spinner's laboratory at the Children's Hospital of Philadelphia has studies ring chromosomes since 2006. Today, thanks to RING 14 it will be possible to **test if the ring structure is responsible for the alteration of gene** utilizing cell lines from mosaic patients who have some cells with the ring and other cells with two normal chromosomes. The scope is to compare these two profiles in order to **find out the causes of the**

seizures and detect more rational treatments. At the same time they will also compare the expression profiles of cell lines of patients with ring which have deletions or duplications with those of patients with deletions or duplications not associated with rings. The Spinner laboratory is focused on ring chromosome 14 (r14) and ring chromosome 20 (r20) but plans to extend their studies to additional rings.

The second grant, financed by RING14 Italy, RING14 USA and Diffusione Tessile

For 10 years RING14 Onlus has promoted and financed scientific projects in the clinical, genetic, pharmacological and rehabilitation fields in order to understand the pathogenesis and detect potential therapies against rare syndromes caused by alterations of chromosome 14. Professor Spinner's research **"Gene expression analysis in the ring 14 syndrome"** represents the second international grant financed by RING14 Onlus, supported by RING14 USA. Since 2012 all the projects financed by RING14 are evaluated and selected on a peer review basis, i.e. by an **international board of scientists** who will also be in charge of checking the progress and the achievement of the set goals. Professor Spinner's study particularly will almost entirely be backed by the charity aid ["Let's shelter children from epilepsy", promoted by Diffusione Tessile and Ring 14 Onlus](#): starting from mid November 2012 up to early January 2013, **12,000 closable umbrellas** (stamped with the wording "Let's shelter children from epilepsy" and the two logo's of Diffusione Tessile and Ring 14) **will be on sale at € 4,00 in 16 shops of the most famous Factory Outlet in Italy.**

"Opening a new front in the scientific research" so Stefania Azzali, Chairwoman of Ring14 Italy, "for us is a reason of hope and happiness. Ever since have we fought in order that rare syndromes be not neglected and our kids may be treated in a rigorous way. We have understood that in order to avoid scientific dispersion and convey all resources towards innovative projects, we need to join together and involve international bodies. This is the target of RING 14 International".

Interview to Dr. Nancy Spinner, Responsible for Research

1. On which assumptions is the genetic research "Analysis of the gene expression of the Ring14 syndrome" based?

We are investigating why ring chromosomes are associated with abnormalities, with special interest in seizures because this phenotype is shared between different ring chromosomes. Our first hypothesis (assumption) is that genes are the ring itself are not expressed normally (that is copies are not made into RNA and protein the same way these genes are when they are on a normal chromosome 14).

2. Which is the main result it aims at?

Our first important result is to determine if gene expression from a ring chromosome is normal or decreased.

3. Which will be the most complex feature to face?

The most challenging part will be first to get the cells with rings to grow well in a culture dish, and then to carry out the analysis of gene expression, which requires many experiments with careful attention to what normal cells we should compare our results to.

Engagement, support, future: the Associazione Internazionale RING 14 Onlus

Founded in 2002 in Reggio Emilia Italy by the initiative of a few families of children affected by a rare genetic disease caused by aberrations of the chromosome 14. For 10 years it has worked rigorously in order to grant support to those who live daily serious disability situations and to promote projects of international scientific research. Engagement, support and future and the values that encourage the RING 14 staff and volunteers. The mission is to mitigate the suffering of children and families and also to write about the story of this devastating and almost unknown pathology, by favouring the development of more and more early diagnoses, by helping discover efficient therapies and strengthening the net of contacts between physicians and people.

Those who wish to support this research may do that at any outlet of Diffusione Tessile or through Associazione Ring14

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