For the first time Telethon is sponsoring a research project in favour of RING14

A new scientific project of Professor Zuffardi of the University of Pavia has obtained the green light:

It's goal is to analyse the function of the missing genes in patients affected by Ring14.

The study will find an important support by the Biobank of Ospedale Galliera of Genova.

The engagement in the scientific research is now stronger. After waiting so many years and after having worked so hard, the **International Association RING14 Onlus** is now receiving a wonderful news: within the frame of their 2013 Call, the Telethon Foundation has sponsored the project "RING 14 SYNDROME: TOWARD A DETAILED GENOTYPE-PHENOTYPE CORRELATION", which is being submitted by **Professor Orsetta Zuffardi, Professor of Genetics at the University of Pavia**, in cooperation with **Professor Nancy Spinner, Director of The Children Hospital of Philadelphia.**

The research **will last 2 years** and will aim at analysing **blood samples and cutaneous fibroblasts** in order to understand the **function of the missing genes** in patients affected by Ring14 and other rare syndromes involving chromosome 14.

"*The main result we expect to obtain*", so **Professor Orsetta Zuffardi, project leader**, "*is to establish a precise correlation between deleted genes and corresponding phenotypic alterations and to show that at the moment it cannot be excluded that the appearance of some symptoms in patients with the ring might be due to alterations of the expression of 14q genes rather than to their deletion".*

The final goal is to identify efficient genic therapies in order to treat the main symptoms like epilepsy and psychomotor retardation and grant a better quality of life to those who live with these rare diseases.

"This Telethon financing" according to **Stefania Azzali, Chairwoman of RING14 Italy**, "*would be a great achievement as well as the confirmation of our efforts after 11 years of activity. Our study has been selected among so many competing, due to the excellence of the project, the high level c.v.'s of our researchers and the reliability of our association in terms of engagement in the research, gathering and disclosure of data. We should not forget either that in 2009, RING14 Onlus started an important agreement with the bio-bank of the Ospedale Galliera of Genova in order to gather biologic samples from families from all over the world which are now invaluable for this project"*

The **Bio-Bank of the Ospedale Galliera Of Genova,** which is part of the **Telethon Genetic Bio-bank Network**, is a centre qualified to **store the genetic material of patients affected by rare syndromes**.

The bio-bank has been founded in order to keep this material at the disposal of the researchers from all over the world, to **develop new studies, to grant a better access to the samples and availability of interesting data**, but also in order to avoid scientific data being uselessly scattered and allow those who are affected by rare syndromes to look at better future.

Interview to Professor Orsetta Zuffardi, Research Leader

1. **Which are the assumptions of the research you are developing ?**
   The knowledge of the Association RING14 Onlus through its Chairwoman Mrs Stefania Azzali and a long experience of studies on the genotype-phenotype correlation in individuals with chromosome abnormalities.
2. **More in detail, what kind of contribution will your experience make ?**
   I will have to determine if within a significant number of cases there are phenotypic differences between individuals with distal deletions 14q and individuals with ring shaped chromosome 14. A preliminary test would show that individuals with ring shaped chromosomes suffer from cyclic epilepsy, whereas individuals with comparable deletions in the distal region of chromosome 14q do not.
3. **Which is the main result you hope to obtain ?**
   I expect to show that a precise correlation between deleted genes and corresponding phenotypic abnormalities exists and that it cannot be excluded that at the moment the appearance of some symptoms in patients with the ring might be due to alterations of the expression of 14q genes rather than to their deletion".
4. **What will be the most complex aspect you will have to face ?**
   The possible expression analysis. Ideally we would need to transform the nucleated cells of the patients' blood in neuronal cells (iPS: induced pluripotent Stem Cells) in order to understand, versus the necessary controls, any possible alterations in the expression patterns. The drawback is not to obtain the neuronal cells, which anyway is not an easy task, but to understand what kind of neurons we need to obtain by means of the iPS methodology.