

Dear volunteers, members and families,

As most of you already know, since a few days I am back from the U.S., where I met our researchers and the U.S.A. Ring 14 Board.

I am very happy and proud to tell you that I have been welcomed with warmth and the people showed a true involvement in our cause.

After two days of relax I spent in Tampa in Silvia Weizman's home, (thanks Silvia for your hospitality!), I moved to Philadelphia to start a tour de force of meetings with our researchers.

The Ring 14 U.S.A. Chairwoman, Yssa, and myself met first [Professor Spinner](#), who works at the Philadelphia Children Hospital, which we are sponsoring with a biennial Grant started in 2012, whose target is to understand the mechanisms of the abnormal functioning of the genes implicated in the chromosome 14 syndromes, particularly deletions and formation of rings.

Professor Spinner wishes to request the bio-bank all our samples and to test them immediately since her aim is to obtain significant data and submit an important project to NIH (the American national body that sponsors medical researches) next November. She is really very much involved in our study, also thanks to the rich collection of biological samples we have managed to gather during the last years, which we deem it important to go on gathering also in the future.

Professor Spinner also said she is ready to contact some neuroscientists in order to achieve the study on the neuronal cells and has envisaged the possibility to create the IPS cells directly in her laboratory. She still remembers [our 2011 WS](#) and congratulated us saying it was the best she had attended. Her studies are aimed at understanding the cell mechanisms causing the symptoms in the Ring 14, particularly epilepsy. Once these results are achieved, and thanks to the creation of the IPS cells and the [Ring14 mouse](#), the new prospects could really open new horizons for the treatment of the single symptoms starting from epilepsy.

Later on Yssa and myself met with [Doctor Anne Cherry](#) of the Boston Children Hospital. Anne said she was very happy of the results achieved by her study, that allowed her to reach the target of re-creating IPS cells with Ring 14 (something that looked quite difficult to obtain).

The most immediate and efficient use of the IPS is in pharmacogenetics. By re-establishing the neuronal cells of every single patient – thanks to the future work of a neuroscientist – creating individual IPS's for each of them, it will be possible to test the drugs in vitro, especially the anti-epileptics and check their efficacy on each individual. This will prevent us (hopefully in a near future) from testing drugs on children, avoiding all the consequences this entails in terms of quality of life, risks and side effects, since we will do it first in vitro. The long-term development of these cells, on the other hand, is linked precisely to the transplantation on the brain tissue of reprogrammed neuronal cells that should replace the abnormal ones. For other kinds of pathologies (cancers or transplants) the researchers are already testing it on mice and have recently started also on the brain. That is why a Ring 14 mouse will be very useful in the future.

My last “scientific” meeting was with Pietro De Camilli at Yale University, where Pietro is the Director of the Programme in Cellular Neuroscience and Neurodegeneration of the Kavli Institute of Neurosciences. He is also the father of a girl affected by Ring14. He is very happy about the way the projects are developing. We have started to outline together the next international WS we will organize in Italy by the end of 2014.

Finally I took a flight to Lafayette, Indiana, to Yssa’s home, where I had the pleasure to get acquainted with Karen and Debbie too, who are as well members of the [RING14 USA](#) board and mothers of kids affected by Ring14. Together we shared our wish to work on common projects though keeping our own individuality while we expect to create, hopefully in the short run, an International Ring 14 that might include and coordinate all the Ring 14 offices that will be opened in the future in the various countries.

Yssa has set up a fantastic charity dinner: there were more than 150 guests, excellent food, very interesting presentations of our projects and a charity fundraising auction, a real success !

When we started 11 years ago, none of us could even imagine that in such a short time we would attain so many significant results. Nobody would have ever dreamed that internationally renowned researchers would be interested in us and appreciate our work, devoting their time and attention to so rare syndromes.

I therefore wish to thank all of you because without your help nothing would have been done. I thank the families for having given the blood samples and the clinical data, I thank our supporters and donors for having believed in us and contributed to the economic sponsorship of our projects, as well as our members and counsellors for their engagement in working for Ring14 and I also thank all the great volunteers – you are more than 200 ! – that help us giving us their precious time, their skills, their energy, the strength that pushes us forward and makes us look at a not too distant future where our children can have a treatment.

A SINCERE THANK FROM THE BOTTOM OF MY HEART: YOUR ARE ALL GREAT PEOPLE !