Il Giornale di Vicenza - 2001

With the "Baschirotto" funds. A researcher supported by the Association illustrates the results obtained in laboratory and gives go ahead to the final trial.

Great announcement, a treatment appears

Metachromatic leukodystrophy: from the condemnation to the first steps of a real change

The discovery is great. It is destined to have unconceivable consequences and to give hope to a lot of families forced to live the dramatic reality of a rare disease which was incurable till yesterday.

Antonella Consiglio, scholarship holder at the Association and researcher in the Claudio Bordignon's team at San Raffaele Hospital in Milan, was thrilled while announcing the news publicly at "Mauro Baschirotto" Association for Rare Diseases.

The metachromatic leukodystrophy therapy trial, financed by" Mauro Baschirotto" Association, has given excellent results. This will lead to the beginning of the toxicologic trial in Primates (monkeys) and then it will be applied in man. Matter of some months, certainly not of years.

Metachromatic Leukodystrophy is a rare disease that destroys the central nervous system cells, leading in in a period of two years for the most serious cases, to a cerebral degeneration, and to the death of the child. Children are the individuals affected by this serious disease.

For many of them, a concrete hope will arise, after years of dark and resignation.

This would be enough to give importance to the discovery in scientific field.

In addition it is the first discovery like that in the world, together with the one in France and USA, carried out with a different method.

There is something else. The discovery implications are different and extremely interesting, as Antonella Consiglio said "Starting from a genetical therapy, that consisted in the direct treatment of the animal's brain (a mouse), precisely the white matter located in the hippocampus, we noticed that only one therapeutic gene injection was able to prevent the disease in the healthy subjects and to cure the subject in which the disease was already arise. It is not all. We noticed that the injection effects lasted even for some months: in animals that normally live two years it is a long lasting period. The enzymatic activity that was absent before, now was operating with a general great surprise".

"The discovery - carries on – was really stunning, the first obtained in the Central Nervous System. Our cleverness was to identify the carrier virus, the lentivirus, that transports the therapeutic gene in the cells. The other great discovery was to observe a gain not only in the inoculated area, but even in the rest of the brain.

The neuronal regeneration operated in areas far from the treated zone, this makes us thinking that this is the right way to greater discoveries".

This aimed that even for many other pathologies soon there would be a change, not only for metachromatic leukodystrophy, whose therapy can prevent the disease in presinthomatic patients (apparently healthy) and to make the disease regress in case of effective disease".

“These incredible results has suggested an open question: why there have been a cellular repair even in the distant cells? Surely there have been a correction in the neuronal damage but we do not know why”.

Now, discovering the vector virus’ efficacy, the step to arrive to other important discoveries is short.

In the USA, where a group of researchers from Sal t University of S. Diego, headed by Fred Gage, are concerned in the regeneration of nervous cells such as in the field of the marrow lesions, these discoveries lead to important results.

From the knowledge of the base mechanisms we could solve unfathomable mysteries.

Giuseppe Baschirotto: extraordinary moment now we must run

San Raffaele and other Institutes thank the society from Vicenza “Without them all the things were stationary”

Associated to names as San Raffeale in Milan, which with Claudio Bordignon was the innovator of genetic therapy, Cardiolo in Turin which with the contribute of Professor Naldini head of the Institute for the rasearch on cancer collaborated to develop the lentivirus vector of the therapeutic gene and the Psychobioloby and Psychopharachology Insitute in Rome where was studied the behaviour of animal that were treated with the new therapy, there is Bird Foundation too.

The Mauro Baschirotto Institute for Rare Diseases was the promoter and supporter, during these years, of the research that lead to the discovery of a therapy capable to fight against metachromatic leukodystrophy, at least in the animals.

Giuseppe Baschirotto was looking for this moment for years. His and his wife, Anna life has completely changed direction after they have lost theyr son, Mauro, due to a rare disease.

His main aim is to give to parents afflicted by the same destiny the answer that he could not have at that time. “my wife and I – narrates Giuseppe – have always believed in gene therapy. This is a special moment for us, even if it must be only the start”. Now we must run more and more faster to save as many child as possible”.

Association for Rare Diseases in Costozza was thanked publicly by all the scientists involved in this discovery in the Nature Medicine's page. A right and sincere act, particoularly for Doctress Consiglio.

But how much was important the role of Baschirotto Association? “I would say essential – underline Antonella Consiglio – without the Association's financing it wuold not be possible to start with the research. Thanks to the Baschirotto Association San Raffaele's equipe headed by Bordignon, the initiator of gene therapy, Cardiolo in Turin which with the contribute of Professor Naldini head of the Institute for the rasearch on cancer collaborated to develop the lentivirus vector of the therapeutic gene and the Psychobioloby and Psychopharachology Insitute in Rome (Cnr) where the behaviour of animal that were treated with the new therapy was studied arrived at this extraordinary result. Scientific field will not thank enought this people”.

Antonio Simeone