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The rare disease. In 2002 the first official recognition.

The discovery born in Vicenza

The treatment for "metachromatic leukodystrophy" found at San Raffaele, after the study at "Baschirotto".

Eugenio Marzotto

Longare

Plays around with small plastic packagings, inside them there are samples of human DNA from Bogotà and La Sapienza University of Rome. On the table there is a piece of paper with Chinese chromosomes inside.

Everything is so normal at the Baschirotto Association in Costozza that you do not think that here they work to cure rare diseases. Millions of hours passed at the microscope to obtain a little piece of scientific success. Than arrived the news that all the efforts have a good result.

The study. The scientific discovery of the year come from San Raffaele hospital in Milan, a researchers' equipe discovered the treatment for the metachromatic leukodystrophy, in Vicenza there is only one case, the prevalence is 1:250000, but it is difficult to estimate.

That discover could not arrive without the contribution of Baschirotto Association that carried on for ten years the research, which passed at Telethon financing new studies.

Giuseppe and Anna Albarello, companions in a lot of adventures, were able to put an essential contribute in the Italian medicine development.

"In 2010 my wife and I were at a conference in Milan where the Telethon head announced that the metachromatic leukodystrophy trial started with an Albanian girl. In that occasion they explained from the stage that without our work in the Association it would not be possible". It was the public reward for the work lasted eight years.

From Calabria. Everything starts in 1992 when a mother from Calabria asked to be helped in Vicenza: "We were at the first steps of genetics – said Baschirotto – talking about rare diseases was a waste of time". The contacts with the guru of genetics started, for example Claudio Bordignon (the first in the word who have completed a gene therapy) who said to Anna and Giuseppe "none could enter in nervous system, find money and researchers, then we will see".

The research financed by the foundation in Vicenza in 1993 moved to San Raffaele and the researches started to fight the mortal syndrome, Antonella Consiglio, Gianmaria Severini and Antonella Sangalli worked hard under the oversight of Bordignon who understood then years before that "purifing" the HIV virus and injecting it in the cells the syndrome would have been cured.

In 2002 the change with the trial with some mouses lasted six months. The prestigious journal Nature wrote that the research obtained a great scientific result because there was the regression of a neurological damage, so it is possible to cure, considering the results in animals.

Is the worldwide sign of the Baschirotto good work.

Second phase. The trial in human occurs, the collaboration with Bordignon stopped and the research passed to Telethon which financed the project.

The first results arrived in 2010.

but that killer-syndrome has killed some children and while the Baschirotto continued to keep in touch with San Raffaele, three researchers find the right way. The successor of Bordignon is Luigi Naldini who, with Maria Sessa and Alessandra Biffi, arrived at the final phase. The project a follow-up of six children aged 3-6 years.

“They opened an essential way, the demonstarted that staminal cells induced by purified HIV can produce a regression of the syndrome” said Anna and Giuseppe.

Even in Vicenza there is a case, Alessandro and his mother Elisabetta who in the book “Hope is a wild flower” wrote: “Everyone has his way to … ”.

in these days the etic commission of the minister is evalutating the stamina method. Giuseppe said: “It is a madness opposing to it”.

What is this?

The Killer -syndrome

Metachromatic leukodystrophy is a serious rare disease, characterized by a progressive accumulation of toxic substance in the central nervous system. It raises often in the late childhood and presents walking difficulties, optic atrophy and motor regression, and finally involved the mental functioning.

Symptoms become serious until a state of decerebration to the death.

New studies

Doctors have isolated the staminal cells from the children's bone marrow from the hip. Then they have operate with gene therapy with HIV virus and finally they have injected it in the children, without rejection problems. The trial started in 2010 in sixteen small patients 6 of them were affected by neurodegenerative disease; after three years three of them were cured.