Noelia Foundation participates in the Conference on Rare and Complex Diseases at the Hospital Sant Joan de Déu in Barcelona

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Last February 27, 2020, the Conference on Rare and Complex Diseases and 1st Exhibition of Patients' Associations was held, organized and hosted by the Hospital de Sant Joan de Déu Research Institute (IRSJD) and the Institute of Bioengineering of Catalonia (IBEC), in collaboration with the Spanish Nanomedicine Platform (NANOMED Spain).

The Institute for Bioengineering of Catalonia (IBEC) is a research centre, based in Barcelona, that carries out interdisciplinary research at the frontier of engineering and life sciences, with the aim of generating new knowledge, integrating fields such as nanomedicine, biophysics, biotechnology, tissue engineering and information technology applications in the field of health. The Spanish Nanomedicine Platform is an initiative that intends to bring together the main Spanish players in research, industry and administration, in order to promote a common strategy in the field of nanomedicine.

For the purposes of clarifying concepts, nanomedicine is understood as the use of nano-elements (elements which size is measured in nanometers, that is, smaller than a cell) such as viruses, proteins or DNA sequences, among others, for the treatment of diseases. This field includes, for instance, gene therapy, the use of viral vectors, the ultra-targeted administration of cancer drugs by lysosomes or the creation of artificial biological organs. Nanomedicine is part of what is considered the medicine of the future, which encompasses artificial intelligence for interpreting results, the application of virtual reality to diagnosis, telemedicine to monitor patients remotely and automatically, robotic orthopedics, or the creation of therapeutic elements using 3D printing. Although these alternatives sound like science fiction, today and tomorrow's reality is nanomedicine, the main trend in new treatments, both in terms of the development of therapies themselves and in the design of vectors that allow therapies (new or current) to reach the target tissues without affecting the rest of the body.

In this field, Spain is the third country in the world ranking of scientific publications in the field of nanomedicine. Barcelona itself is up there too, holding the 5th position among world cities, ahead of New York, London, or Bethesda (Maryland, USA), home of the US National Institutes of Health (NIH).

As regards the role of the Sant Joan de Déu University Hospital in the field of rare diseases, to date, it has over 17,000 patients with rare diseases registered, which represents a sizeable 7% of all consultations. However, if we look at the patients under the care of the Neuromuscular Diseases Unit who participate in any of the clinical trials in which the hospital is involved, the percentage rises to a 20%, evidencing the high level of current research activity in this field. And this is not the case only at the local level, but also globally, in 2019 over 200 clinical trials testing new gene therapies were authorized. And this number has been growing exponentially, year after year, which shows that, very soon, this type of therapies will be part of regular clinical practice.

Leading researchers from various fields of health care participated in the conference itself, all with the common denominator of either participating in the development or being potential beneficiaries of the use of nanomedicine for the treatment of the diseases they work on, such as sudden death, cystic fibrosis,

amyotrophic lateral sclerosis (ALS), muscular dystrophy or something as common as diabetes. Specifically, we had Dr. Georgia Sarquella-Brugada, an expert in arrhythmias and sudden death and an IRSJD researcher who spoke about developmental cardiovascular diseases; Dr. Andrés Nascimento, a child neurologist at the Hospital and a researcher for the IRSJD, who addressed the issue of applied research in neuromuscular diseases; and Dr. Josep Jiménez Chillarón, an IRSJD researcher who spoke about metabolic diseases in children.

In this exceptional framework, and as the only representative of patient associations, Noelia Foundation had the opportunity to present its project, given the relevant role it plays and the direct impact it generates, despite its modest size, in the advance of research in Collagen VI Congenital Muscular Dystrophy.

In their speeches and across all presentations, the speakers highlighted the crucial role nanomedicine plays (and will play much more in the near future) in the development of new therapies and, in particular, of treatments for genetic diseases, whether they are rare or common. At a global level, the application of nanotechnology to medical therapies, whether genetic or pharmacological, offers advantages in several areas. First, it allows designing vectors that protect the therapeutic agent (be it a drug, a protein or a DNA fragment) from degradation during its transit through the bloodstream. Also, encapsulating the therapy reduces, or may even prevent, systemic toxicity as it isolates the agent from the rest of the body. Finally, the molecular design of, either the therapeutic agent or the vehicle (virus, nanovesicle, etc.) may optimize the affinity of the treatment for the target tissue, which increases the concentration in the target cells and, thus, the effectiveness of the treatment.

Focusing on our particular area of interest, Dr. Andrés Nascimiento explained that, following the trend of previous years, in 2019, 95 new neuromuscular diseases have been described, reaching a current total of 1,042. Fortunately, most of them are genetically characterized, which is the first step in the development of any kind of treatment.

"Thanks to the new technologies and nanomedicine," says Dr. Nascimiento, "more and more light is being shed on the nature of rare diseases and their treatment alternatives". As an example, he points out that, recently, a therapy for Spinal Muscular Atrophy (SMA) has emerged, based on the administration of oligonucleotides (nanoelements). This new approach manages to reverse the clinical symptoms in young patients, and even preventing their appearance if applied in the neonatal stages. In the case of neuromuscular diseases, unfortunately, the target of therapy is not the cerebrospinal fluid as in SMA, which is very a contained environment, but the muscles, which are distributed throughout the body. This implies that the therapy has to be distributed in a systemic way with the difficulties this entails, as regards of potential side effects and the level of drug that effectively reaches the muscle. It is precisely in this area where nanomedicine may contribute most, both at the therapeutic level, thanks to the design of specific vectors of the target tissue, and at the research level, through the creation of artificial models of extracellular matrix from patient cells, which help predict the effect of different therapies in the laboratory.

In conclusion, this brief but interesting conference stressed the relevance of nanomedicine in the present and future of therapeutics. It also shows that, without having to travel far away, we can count on a pool of local research groups that are achieving very promising results. At the speed at which new therapies are being developed, it will undoubtedly not be long before we have new options to help patients with rare diseases.