

Hello everyone!

This quarter has been intense for the Niemann-Pick Foundation of Spain. As you know, during February we were focused on National Niemann-Pick Day (February 17th) and on the World Rare Disease Day (February 28th). On the World Rare Disease Day, we collaborate and participate in a lot of activities that were organized nationally and internationally. It's very important for all of us to be able to keep in touch with entities that work with rare diseases to know the news, the treatments and the trials that are carried out to improve the quality of life of those affected by Niemann-Pick. and ASMD.



During the World Rare Disease Day, Julia González, our family coordinator and legal representative, and Manolo Campos, patron of our Foundation, attended the main act of the event in Lleida, organized by FEDER and the Platform "Malalties Minoritàries" from Catalonia.



In March, we have already begun to prepare our XXII Congress, but we have continued to participate in more activities, such as, for example, attending the Institutional act for the World Day of Rare Diseases in Andalucía in the Andalusian Parliament, which was attended by patron and delegate in Sevilla, Juana Contreras.

Also, in March, we participated in a webinar organized by the ASMD Spain about "The importance of Patient Registries in rare diseases" with Dra. Dolores Ledesma and Dr. Jesús Villarubia.

In April, we participated, in person, in the Congress of Orphan Drugs and Rare Diseases 2022, in Sevilla. It was a highly prestigious congress, with the presence of many professionals, in which they discussed promoting research and achieving full equity in access to diagnosis and treatment by 2030, as the main challenges in the fight against diseases rare.

Also, in April we commemorate the World Day of People Without Diagnosis. We join the campaign "Without diagnosis, there is no prognosis" because an early diagnosis is essential to know what a patient is facing.



The main act of this quarter for the Foundation has been our XXII International Scientific-Family Congress. This year we have done it in a hybrid format, face-to-face in Barcelona, and in virtual mode for all those participants who connected online. It was a very productive congress, where we were able to count on the most prestigious professionals and researchers who are currently dealing with Niemann-Pick and ASMD. We are talking about advances in the disease, about new

therapies, new trials, and future projects for those affected by the disease. In addition, dedicated Saturday afternoon to learning more about how the INPDA and the INPDR work.



The Congress gave us the possibility for most of us to meet again in person after two years of the pandemic. It was very exciting to hug each other again and see each other after so many months without being able to do it. We left the Congress with renewed energy, with challenges ahead, and with a lot of work to do.



We have very important news to give you: on May 25th, finally, we will start the clinical trial with Efavirenz! After having to delay it twice due to COVID19, it was time to start. During May, five patients with Niemann-Pick type C started the tests. The trial is being carried out at the Hospital Universitari de Bellvitge, in l'Hospitalet de Llobregat (Barcelona), and is led by Dr. Jordi Gascón and Dr. Sebastià Videla.

In the next few days, the rest of the participants in the trial will start the tests. The approximate timing is 52 weeks with 6 control visits per patient. We have a lot of hope for this trial, and we hope that the results will be as expected by all. We will keep you informed!



After the Congress, in June we also have two events: we are going to participate in the Assembly of members of FEDER (Spanish Federation of Rare Entities), of which we are a full member. It is a very important event, because FEDER is the muscle of the associative movement of rare diseases in Spain. A part of this event, on June 26th, in La Unión (Murcia) we are going to do a benefic bingo. It will be organized by the family that lives in La Unión, and this year will be the second edition. Thank you, from here, to all the people who with their generosity are going to make this charity event possible to help all those affected by Niemann-Pick and by ASMD.

We also inform you that our Foundation directive was changed: the secretary of the foundation was renewed, and two new patrons entered to replace two more. From here, we would like to thank Cristóbal Fernández, secretary until this year, for those 22 years of dedication to our Foundation. We also appreciate the dedication of Mari Cruz Rubio, who has left her position on the directive but continues as a beneficiary family of the Foundation. And we make a big welcome to Carmelo Fernández, Javier Galera and Carmen Calatayud as new members of the directive Foundation. The new secretary is José María Fernández, who was previously a patron. From here we also wish him good luck in the position.

We wish you all a happy summer, and we'll read you in the next newsletter!

