

Good morning!

I am Elena, president of the Spanish association of ceroid lipofuscinosis, I am going to tell you how I got to be here.

The 18th January 200 I heard for the first time that word ceroid lipofuscinosis. It was in the hospital nino Jesus de Madrid. It was the diagnosis that was being given to my daughter Paula, a beautiful girl who by the way was only eight years old. In that moment, my life changed forever. Almost instantly a lot of questions started to form in my mind when I heard about the blindness, madness, ataxia, epilepsy death, death, death, NO, not that, that is impossible, there must be a mistake, that cannot be happening to my daughter, that doctor doesn't know what she is saying, it's my little girl; why is that happening to her? She is only a girl, my girl.

I don't really remember the next two days at the hospital. Those were like days lived in a kind of cloud of bad thoughts, I couldn't hear nor see anybody, I only remember that constantly lots of bad questions were in my head; what have I done wrong, where have I made mistakes, what mistake could I have made and over and over again I asked the doctor are you sure of the diagnosis, couldn't it be a mistake, is there any place where my girl can be cured.

When we came back home we arrived with a feeling of defeat, loneliness and failure. The days were unbearable but the nights were even worse, in my head were lots of images that I was trying to erase, but that once again came back to dominate my mind.

It was then when I started to think that it was only a nightmare, a terrible nightmare that it was only going to disappear with the light of a new day. But the next day everything was still the same. I slowly approached Paula's bed and I saw her so beautiful and defenceless and so full of life that I couldn't understand anything.

After a while we started a frenzied search for solutions, we had to do something, somewhere there had to be someone who could cure my daughter. We asked for another diagnosis from another neurologist and another and another, but all said the same. I started to think to take her to the United States to London, to Paris or any other place in the world where something could be done for this type of patient. But all the information we got made us realise that there weren't any successful treatments for my daughter.

I think that until here my story is the same as all of you have lived.

Afterwards I started to feel like a weird bug, it seemed like my daughter was the only one in the world with this illness, no one knew about this damn sickness even doctors made weird faces when I told them about this strange word. Day and night I looked for other families with ill children but I couldn't find anyone in Spain.

Through the internet I contacted some South American families and the association of BDSRA who gave me information and a shoulder to cry on. It was then that I realised

the importance of being in contact with people who are suffering the same as you, who have the same doubts and who get peace listening to other people with the same problem.

With time I started to keep in touch with a Spanish family and I felt a great sense of relief. At this point an idea came into my head, to have a group of families in Spain but we were just a few, and we had to stay next to our children 24 hours a day. In spite of this, the idea was still there, I thought that even though we were just a few families we needed a meeting point, I considered that the strongest association was BDSRA and that it could be our guide which would inform us and give us instructions of how to keep moving in this difficult process.

So I contacted them and after several months on the 23rd February 2006 we finally got to become a legally constituted delegation of the BDSRA.

During this time we had found new families who came with a need for information but also with a need to share experiences and shoulders to cry on.

The bad thing is, that when children die the majority of families stop keeping in touch with the association because after losing their children it is very painful for them to carry on being in contact with this illness.

The Spanish association I represent is very small, it is formed of 15 families across the whole of Spain with little free time to be able to work within the association.

During this time we have got together on two occasions, in our first meeting we had the neuro paediatrician expert in Spain, Dr Pineda, and in our second meeting we had doctor Ines de Halac, an Argentinean scientist working on this illness.

The fundamental work that we are doing in the association is the practical exchange of information between families, either about help they can obtain from the government, or about their children will progress and about the medications given to their children.

As we have low participation and a small number of members we cannot hold many events. We normally pass the information we find to others, and when a family is worried about a new manifestation of the illness, they can ask other families who have already experienced that manifestation in their child.

For this reason we come to this conference with the intention to belong to a unique federation where combining our efforts and resources, is the way to achieve our objectives.

- That governments get involved with the illness and that specialist hospitals are created at a world wide level, as well as getting politicians from Brussels to have in their programmes and health policies specific measures directed towards this illness.

- To incentives the scientific community to start new studies or to continue those ones already active. Promotion of investigation of Battens Disease.
- Promotion of sponsorship so that researchers can undertake research in laboratories of universities anywhere in the world.

The new biotechnology advances, especially in genetics, give us hope that it will be translated as soon as possible into clinical treatments.

- To get laboratories involved with the illness even though it is not attractive to them to investigate it.
- To promote the knowledge of ceroid lipofuscinosis for health professionals and society in general.
- To establish a statistical basis and real data of the situation of children with ceroid lipofuscinosis in every country and that this data can be consulted by all those who are interested, of course without providing their real names and ensuring data protection.
- That all patients can be considered for clinical trials and that they are not disadvantaged because they are not in the country where the study is being conducted.
- That all patients can use the public health systems to conduct genetic tests and therefore identify this pathology and avoid its hereditary transmission.
- To promote the specific knowledge of Battens Disease in health professionals such as Paediatricians, Neurologists, Psychologists, Ophthalmologists, Physiotherapists and any professional who has to treat our children, with the purpose of detecting this type of illness promptly and avoiding delays in referring the patient to the correct specialist for its diagnosis. The average delay in its diagnosis is five years, from the onset of the first symptoms to a correct diagnosis. The primary action must be to guarantee an accurate diagnosis, treatment and appropriate support for the affected family.
- New common guidelines so that teachers can treat our children appropriately. To create a unique manual to inform schools so that this manual can be used as a guide.
- I believe that it is important to belong to EURORDIS, the bigger we are the better we can fight this illness.

Each time the families suffering is more evident.

All the problems have the same origin: the lack of knowledge of the illness by many

professionals, the lack of communication between professionals and of course the lack of research.

As EURODIS says, we will recognise the importance of joining forces on an international level to:

- Exchange information about the illness.
- Give and receive advice and support.
- To combine resources.
- To expand the capacity to raise funds and to have access to economic resources at a European level.
- To have a stronger voice to protect our illness in front of world wide institutions.
- To promote the research on ceroid lipofucinosi and to support research projects.
- To cooperate with doctors and researchers. The connection between patients is capable of improving the cooperation between professionals.
- To work towards common protocols for Battens disease.
- To organise ourselves to be able to participate in clinical studies and research.
- To be in a stronger position to negotiate with industry.
- To support other groups of patients in less developed countries.
- To develop tools of common communication and a website.
- To create common projects.

Of course every one of us has part of the responsibility to achieve all of this. We cannot rely on others. Therefore the Spanish association promises to work effectively on all the proposals we get from here.

Thankyou very much.