OSCAR DHOOGE Y SANDRO GORDO
INFO@OSCARDHOOGE.COM - 608372147
AGORDOPRIETO@GMAIL.COM - 639254315

RARELY UNIQUE

The 28th February marks the International Rare Disease Day, the perfect time to reflect by approaching the silent but constant struggle of two rare families, whose time and effort focus on providing the highest possible quality of life for their children. Everything for Adrián and Aina, two rarely unique people.

At present there are about 7,000 rare genetic diseases affecting more than 3 million people in Spain. And around 350 million people worldwide. 5% of these diseases manifest since childhood, while most of them appear during adulthood. In all cases, patients' quality of life is seriously compromised by the lack of knowledge about the disease, the limited access to health services and the partial or complete loss of autonomy.

Suffering from a rare disease implies an unexpected obstacle course constantly undermined by social cutbacks. There are thousands of families facing misdiagnosis, surgeries, medication, and regular visits to hospitals and orthopaedic centres. But, certainly, the most painful thing of all is to see a child growing with restraints, isolated and marked out as being different by the society and public institutions. Lack of knowledge, investment in research and institutional support are the main causes of the marginalization of this collective.

AINA'S SWEET BATTLE

Aina is a lively six year old little fighter affected by a syndrome called CDG (Congenital Disorders of Glycosylation). She lives with her parents and her twin sister, who does not suffer from CDG, in Premiá de Mar (Catalonia). She is one of the 69 patients diagnosed in Spain and one of the 900 known cases worldwide.

CDG is a disease of metabolic origin caused by defective addition of sugar to proteins, an essential process for the proper development and functioning of all vital organs, such as the liver, the kidneys or the heart. People affected by this disease are highly dependent since they suffer from severe mobility and speech disorders, as well as other symptoms such as strabismus or blood clotting and immune system problems.

Despite her young age, Aina is an example of perseverance and tenacity. She attends exhausting school days in a public school, just as the other children do. In addition, she must frequently visit Sant Joan de Déu and Vall d'Hebron hospitals in Barcelona, as well as going to physiotherapy and speech therapy sessions at the Institut Guttmann of Badalona. However, she never loses her smile or strength to overcome every challenge.

María and Joan, Aina's parents, accompany her in all her routines. They combine their daily work with the constant assistance to their daughter's needs. To this must be added their struggle with the CDG Spanish Association, a meeting point for families and affected people with the aim of giving visibility, seeking support and finding solutions to improve their quality of life.

Aina and her family have shown that, with the appropriate resources and effort, CDG patients can achieve significant improvements and integrate in the most of social situations.

THE RARE CHILD LOTTERY

"Lady, you won the lottery. There are only eight cases in the world like this one". Those were the doctors' words shortly after Adrián's birth and that his mother, Mª Elena Prieto, will never forget.

Adrián suffers from Stüve Wiedemann Syndrome, a congenital bone dysplasia characterized by short stature, curvature of the long bones and hyperthermia. Although he was given a short life expectancy, he has just reached the age of majority. He has been recognized as having a disability of more than 65% and, even though his normal mental development, his reduced mobility forces him to depend on his parents to move and to receive basic hygiene.

Since his birth, Adrián has undergone over 20 surgeries to alleviate the symptoms of his disease. The last and most aggressive of them was carried out in July 2012 at La Fe University Hospital of Valencia. To avoid possible paraplegia, doctors had to practice a widening of his spinal cord, forcing Adrián to be immobilized with a halo ring and vest for more than six months, being unable to move his neck an inch.

Besides his physical limitations we must add the social integration difficulties that specially appear during adolescence. "At high school, he was very quiet and hardly related to his classmates. We hope that he will be able to create his own circle of friends at university," says his mother, Mª Elena.

2014 was a special year for Adrián. After three years of procedures with the social services of Valencia government, he managed to obtain an electric wheelchair. On the other side, Adrián began a new stage in his life highly motivated. He was admitted to the University of Alicante, where he is currently studying a Degree in Multimedia Engineering. Thus Adrián, aged 18, has managed to take his first steps towards independence, without the protection of his family and becoming a self-sufficient individual at the education centre. A slow and difficult process in which actions like leaving home, getting on the bus, taking notes or using the toilet, which would seem to be small obstacles, become major barriers for a person with functional diversity as Adrián.

BEING RARE IN A COUNTRY IN CRISIS

The aids that people affected by rare diseases demand in Spain, are highlighted in the study Estudio EnSERio 2 (FEDER - Spanish Federation of Rare Diseases). Advances to obtain an early diagnosis, further support for biomedical research, and specialized professional services in rare diseases and medical home support are needed. Not only financial support, but also human.

However, scientific research is one of the activities most affected by cutbacks in countries in crisis. In the last five years, the Spanish government has reduced by 40% funding for R & D & I (research, development and innovation). This implies a severe blow to the functioning of research institutes and hospitals, causing the mass exodus of scientists and forcing to close lines of research due to lack of resources. This is the case of Sant Joan de Déu Hospital, one of the reference centres in the study of rare diseases of genetic origin.

Additionally, aids for dependent people are becoming even scarcer. Desired improvements are promised while, at the same time, the budget for dependency is being cut back, medicine coverage is being suppressed and household expenditure increases. On the other hand, loneliness, marginalization and abandonment of affected people and their families evidence that the development of activities that promote educational integration of children with disabilities is an unresolved matter for the education system. With the deterioration of the economic situation and budgetary adjustments for dependency, an increasingly number of families wanders alone in their search for information and treatment. Still, they will always hope to find a solution for these unknown and unpredictable diseases.

RARELY UNIQUE

The "Rarely unique" photo documentary project aims to boost the visibility of rare diseases by raising awareness of the need to promote scientific and medical research, as well as human support for these "unique" diseases, which together affect more than 7% of the world's population.

AINA'S SWEET BATTLE - PHOTOGRAPHY BY OSCAR DHOOGE

THE RARE CHILD LOTTERY - PHOTOGRAPHY BY SANDRO GORDO