Every year, the 28th of February (29th on leap years), known as ”Rare Disease Day”, is dedicated to raising awareness on rare diseases. In Europe, one disease is characterized as rare when it affects less than 1 in 2,000 people. Many of these diseases are complex, chronic and degenerative thus causing serious disabilities. Currently, it is said that, worldwide, more than 300 million people are diagnosed with one of 6,000 to 8,000 different rare diseases in existence, 50% accounting for children, from which 30% perish until the age of 5 years old. A very big issue is that usually, these diseases remain undiagnosed for years, or misdiagnosed, leading to a "Diagnostic Odyssey" for patients, while for 95% of them there is no medical treatment. Consequently, while each of these diseases is rare, if we approach it cumulatively, we get a better grasp of the magnitude of matter in hand. Rare disease is something that concerns many more people than we can possibly imagine. In this issue of Nutrischool News we host an interview with a "rare" mom which has a wonderful "rare" eldest son and a very strong family.

Marily, can you tell us a few words about the diagnosis of your son? At what age and how fast did it occur?

Alexander was diagnosed at the age of two years old when, following a long hospital stay, his gross and fine motor skills did not return to the levels that had been previously achieved within the 6-month period that the doctors had set as a reasonable margin. The diagnostic procedure in Greece was slow and ultimately ineffective due to the lack of both appropriate diagnostic – laboratory tools and the shortage of medical expertise in the diagnosis of such a rare chromosomal anomaly.

What were your first thoughts?

The diagnostic answer came from abroad in a short time after our visit, via email, briefly and facelessly. Their analysis, coded in a series of numbers and letters, and their categorization, in a very rare chromosomal anomaly, that affects today less than 100 people in the world. The text ended with the phrase "Do not stand on the diagnosis alone, but rather, proceed by taking into account the potential of the child "… that is, in other words, your instincts as parents. The first thoughts that come to mind are questions that you set to yourself, such as, why did this happen to me? Why couldn't it be diagnosed on the pregnancy tests? After overcoming this stage my husband and I, as technocrats, started searching medical journals and the internet for relevant bibliography concerning the rare condition of our son. No answers were found because this condition had only very recently been diagnosed by the medical group of geneticists and the results of their research had not been published.

We are talking about a rare disease. How easy was it to organize your daily routines and cover the needs of the child? Is there any state support?

The daily routine of Alexander was, and is, a continuous effort to autonomize and smoothen his integration both inside and outside of the family in social structures such as school,

groups of children and adolescents, sports and society as a whole, which unfortunately hasn't managed to overcome mentalities and prejudices of the past and accept and induct differentiation as normal, wherever it may stem from. The daily organization of the child is a collective effort of the family in continuous cooperation with therapists, special needs educators and gymnasts, along with annual medical monitoring. The monthly economic cost of the whole effort is much higher than the student fees at the best private school in our country. State support covers a very small portion of the needs of the child and many times greatly delays compensation of the treatments.

What were the reactions of the family in its entirety and those of your social circle? There is a saying that "true friends become apparent under hardships (in times of need)" what do you believe?

The family from the beginning was very supportive and especially my father-in-law has been dedicated to the progress and future of the child. The siblings have been aware of Alexander's situation since the beginning and treat him equally without caressing him. In my opinion, as in all the difficulties of life a true friend is someone who can truly stand beside you, and Alexander has presented, and remains, an opportunity for our family to have beside us dynamic, and free from Greek society's stereotypes, friends with an intellectual view and open mind going forward. Surely, over time we had a sorting out of friends and relatives because each person has to withstand that which he/she can tolerate.

Did your experience as a "rare" mother influence you in your decision to extend your family?

To me every mother is "rare" because she has to grow, improvising in balance between emotion and logic, children with different characters, inclinations and temperament. In our decision to extend our family pertaining to my little son who has ten years difference from Alexander we were more concerned about examinations and tests during pregnancy in collaboration abroad and not in our decision to extend our family further. Our daughter, which arrived one year after Alexander, posed no concern since we had no idea of Alexander's condition at the time.

You have managed as a family to become a resilient team. Would you like to share with us any tips?

You mentioned the ideal term, "team". I believe that in order to achieve our goals and dreams, despite the struggles that come along in life, while maintaining our integrity and emotional balance we need teamwork and to be open to expand our team with friends that accept and support us. At the same time, outreach should be the sufficient condition under which we will manage to, maybe at some point, achieve empathy in society and eliminate negative sentiments such as sadness and pity.

Is there an institution that can stand beside "rare" families?

Children with rare diseases are encompassed to groups of people with disabilities. Each disease has its own complexities. In the case of our son, we place great emphasis on his socialization and autonomy. Indicatively, I can mention the Scientific link for custom activities - 'Victor Ardant' Association - "NIKI", that was founded in 2000 and supports parents / guardians and children. Also the Structure of Teenagers & Youngsters "Friends", of the civil non-profit company "Timely Intervention", which is aimed at older children, teenagers and young people help a lot. Through these structures, we try to help children become autonomous and self-serving to the maximum extent they can. Moreover, they form friendships, occupy themselves with various, sports and non-sports, activities and are not isolated. The operation of all these institutions is of great importance, because we must not forget that they provide a place where the children can evolve while simultaneously providing assistance to parents. Patience and teamwork is needed above all.

Thank you very much Marily, our team at www.nutrischool.gr wishes you the best for your truly rare family.