Learn about PI3Kδ (APDS) Activation syndrome
Sara's story

Hello everyone, my name is Sara and I'd like to tell you my story. **It's about bravery, love and resilience which shaped our family during the last years.** I was born after a full-term pregnancy, but my entrance into the world was marked by complications that would have triggered concerns, aggravated by the possibility of a partial heart attack in the placenta.

In the first months of my life, after all I was fine. But, starting from the age of two, I began suffering from mucopurulent otitis, a very trublesome disorder that involved ears and eyes, that often made me ill. In spite of the taking of antibiotics prescribed by my pediatrician, my problem was still present, so I went to the hospital for further follow ups. Later I got a very aggressive stomatitis and I needed to be hospitalized.

In the meantime the follow ups and the several examinations highlighted a lymphocytes' deficiency, that explained the frequent otitis and the eye issues. I began to take immunoglobulins intravenous every 15 days to provide for this lack and, in that period, I also was subjected to adenoids' removal. **In spite of challenges and obstacles about my health, I was able to lead a relatively normal life, attending school and often travelling.** The only waiver was swimming.

At the age of 18, I found out to have my first Hodgkin's lymphoma, followed by other two during the next years. At the beginning this disease had been identified as an unknown lymphomatous form, but later, thanks to the cooperation of Centres with branches in other european Countries, **the tests led to the suspicion of the APDS syndrome, caused by a genetic mutation de novo. Although chemiotherapy and issues tied to the lymphoma, I never lost resolution.**

After a decade of a relative stability and a switch to a subcutaneous therapy, I had to face a second lymphoma. Now, after other 5 years, I'm facing a third one, but this time the treatment has been tailored to my conditions. The medical team assessed different options, stem cells transplant included, but the final choice was a monoclonal antibodies therapy every 3 weeks.

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Although, unlike chemotherapy, the response is not immediate, **I'm facing this situation with optimism.** The total prevention of lymphomas is not possible but, thanks to recurring follow-ups, I try to manage my condition at its best. I I often have to go to the hospital for routine or extra controls, like PET, TAC or coloscopies.

Even later my path has never been without obstacles. Today, at 36, I think about my life and the faced challenges. The early menopause, due to chemiotherapies, is an issue often overlooked by doctors. My mental health has been put to a hard test and, although a private psychological support, I'm going on fighting with permanent anxiety relevant to the fact that my body could let me down at any moment.

My hometown hospital and, in particular, my immunologist, played an important role in understanding and managing my disease. I was able to live like my peers, to get a driving license, to attend university, to be indipendent. When I was a child I remember my parents very concerned, going from one doctor to another without having replies. **We fought together: this has been our strenght against a serious chronic disease. I never gave up** and certainly this influenced very much the person I'm today.

During the days in the hospital I met many other patients with Primary Immunodeficiencies; however I 've never recognized myself in their stories. Then, one day, by chance, I found one of the leaflets made by AIP: it talked about the APDS syndrome and finally I recognized myself in the words I was reading. The information collection, the involvement in their initiatives, the meetings with doctors and patients have been source of support and understanding in a path which normally was isolating.

The strenght I found in myself and my mother's resolution are a proof that it's possible to overcome the most difficult trials. I hope that our story could inspire those who face similar challenges and could show that, even in the darkest moments, the light of love can shine intensely.