



Learn about PI3K δ (APDS) Activation syndrome Leo's story

My name is Leo and I suffer from a very rare disease called PI3K δ Activation syndrome, called more simply APDS. I was diagnosed when I was 3 years old; now I'm 20 and I've got a part time job.

When I was a child, I had a stunted growth, for that reason our pediatrician suggested my parents to get me tested for celiac disease. The outcomes showed an extremely low number of immunoglobulines and I was diagnosed a Primary Immunodeficiency.

Further tests showed I had very low levels of antibodies called IgG, IgA and IgE and too high levels of M (IgM) immunoglobuline. **The outcomes of my tests have been sent to labs and hospitals all over the world with the hope to get a definitive diagnoses.**

My main symptoms tend to affect intestine and lungs: these are frequent and disabling. **The treatment I'm taking is called immunoglobuline, that means that I get antibodies every three weeks.** During the years I underwent many tests and treatments: too many to be counted or mentioned.

When I was a child my mother bribed me with little prizes, candies or new toys, every time I was able to face with no tantrum a new follow-up or an umpteenth treatment. This allowed me to be distracted and braver. It sounds stupid but it worked.

I tried in many ways to lead a normal life, managing school, job, friends and medical follow-ups. I was often forced to live in an hospital for months. I'd have liked to go on studying, attending university, but my health got the better of me and I preferred to look for a job to be independent from my parents. Now I'm a waiter in a pizza restaurant. My health condition can leave me exhausted, drained, both mentally and physically. **However, I do my best to perform my role and to comply with the shifts they give me.**





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I'm the only person in my family to have APDS, that means that it wasn't transmitted genetically. Growing up with this disease made me feel alone, I felt like I couldn't talk to anyone about it. Even my sentimental life suffered about it: I felt it was wrong to give the burden of my suffering to another person.

Not many years ago, thanks to a Facebook group, **I found that there are other people affected and this makes me feel less isolated.** Since then I make an effort to tell my condition to friends and relatives, to express what I feel and to share the distresses. I explain my disease to the others in the simplest way possible: I tell them I have no anticorps so I'm inclined to contract many infections.

I'd like to meet in person other people with my same illness, to build a sort of community where we can talk and compare ourselves with each other.

I wish myself not to be stopped by the barriers tied to my illness and to be able to live my future to the fullest.

