



## Learn about PI3K $\delta$ (APDS) Activation syndrome Lucia and her father Carlo's story

My name is Carlo and I'm Lucia's dad.

My daughter suffers from a rare illness involving her immune system, the APDS syndrome. **It took 19 years to receive a definite diagnosis.** Now she is 24 and works as a secretary.

For many years she has been dealing with endless coughings and colds and she often had to be absent from school due to her infections. Every two weeks we had to go to the doctor to get a prescription for antibiotics. She was afraid to be with her peers and to play with them as she feared to contract flu, with severe consequences on breathing and hearing. She gave up with birthday parties and with the afternoons in the parish, in order to defend her health, always very weak. As you can imagine, **this pathology had a strong impact on her social and relational life.**

One day my doctor prescribed me some blood tests to verify possible reasons of a persistent cough. The outcomes revealed I got a pulmonary disease and white blood cell deficiency. The doctor informed me that this deficiency could be inherited and, following his suggestion, I had my three little girls take the tests: two of them were found to have a defective gene.

Unfortunately the final APDS diagnoses still required time and patience, also due to an unsuitable communication between doctors and hospitals. For many years Lucia has been suffering from a general illness, temperature and various infections; on some days she was so sick not to be able to get out from home.

**Since when the illness has been identified and finally the suitable treatment has been adopted, her condition improved and increasingly under control.** Although some infections that cannot be prevented by drugs still happen, today my daughter often feels tired, perhaps due to the many infections contracted during the years.

The life style of all my family had to be adjusted: **we started to pay attention in following a balanced diet and to adopt behaviours that support Lucia and her sister's health.**





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Since the time of diagnoses, however, there was no shortage of difficult days. Our family repeatedly deals with many obstacles and one of these is surely the lack of training of the medical staff and healthcare workers involved in my daughter's illness. It's very frustrating having to explain symptoms and conditions to those who should know about them most and should be able to address us to a right path, above all as we fear the impact that a wrong prescription could have on the treatment and the therapy.

**I wish that there will be more and more training and divulgation about rare diseases such as APDS**, not only for immunologists and other medical specialists, but also for paediatricians, general practitioners and all health professionals.

