



Imagine carrying a 30 kilogram (66 lb) backpack.

If you're a typical person, your legs will probably handle the effort, stiff as they might become. Now picture this in your everyday life. If you're a college student and you should hurry up to deliver that paper to the teacher's office; if you're a kid on the pool, floating and swimming serenely, until the swim coach calls everyone out and you simply can't lift yourself up from the poolside; that backpack goes wherever you go. It's forever your friend. You go to the park for a picnic and everyone sits on the grass - you think twice. Backup plans rush in your mind. Is there someone strong who can lift me up? Is it polite to ask people to lift me up? Is the bathroom close enough? What if it rains and everyone vanishes by instinct? What if world leaders are secret agents from outer space? Wait...

Of course, there are other social conventions that seem to grow incompatible through time. Those cushy and embracing (and lower seated) couches your cousin bought and offered you to sit on. There's always that hesitation in every room you enter and place you visit, that moment when everyone is making themselves comfortable and you're still inside your head processing stuff.

Now imagine you not only wear a backpack, but a full bodysuit with lots of straps and pouches. Each year you add 1 kg to each leg, each arm and your backpack. This should feel like some Shaolin hero special training where you get superhuman strength over time. It doesn't.

The weakness builds up, so it comes a time when you walk a little wobbly. If you go through a crowd, the fear of bumping on someone is the risk of falling gently, slowmotionly, inexorably to meet the floor. Everyone's like "what the hell just happened". Some of them half crack a laugh until you explain you're handicapped and need to be lifted which makes everyone awkwardly polite and apologetic and you make yourself watch it from the clouds and take notes for a standup comedy later. But you work on it. You adapt. You manage and improve your momentum at top efficiency at which point Maranello engineers start giving you phone calls to ask you for advice on physics. And there's the terrific friends that want to make you belong and go like "hey, let's go to the stadium cheer for [insert favorite team]!". You tag along and, behold, there are only high steps and low seats. *facepalm*



But really, there are a lot of cool things about having Pompe. Besides the naked-slip-in-the-cold-bathroom-floor and messy-toilet-seat episodes you'll have to entertain your peeps at the bar, you can petition exclusive parking space and you can always explain to your unfunny friend that you didn't laugh at his joke because you have face paralysis (true thing, yo). Oh, and if you didn't have eye lid surgery, you can sleep through class and boring couples reunions and pass along as being awake ("I have a hard time lifting my eyelids, guys. No joke!"... Avoiding snorts, however, is a technique of its own... On the Pompe email lists we share and improve these life-changing tricks between world domination plots).



My doctor told me to quit the dark side. I'm using patches to handle the withdrawal...

I guess this is a good look at how my Pompe lifestyle feels like. I'll fill in below with some medical details for the technically minded folk.

Since childhood I had unconventional but otherwise accepted weaknesses. They were regarded as a child's expected particularities. The context was such that inhibition and lack of information always contributed to mask my condition.

At 5 years old, the most evident symptoms were mild neck and abdominal weakness (being unable to lift my head and torso from a laid back position) and lower than average self-lifting power; on occasion where the children played physical games, the discrepancies were evident. Social awkwardness was, in my mind, the frequent reason to hide my symptoms (the sustained strains on my muscle tissue so I could fit in and do what everyone did only grew me at odds over time).

By age 12, I developed ptosis on my right eyelid. I soon had cosmetic surgery.

I was generally able to mitigate the overall symptoms by being motivated in sports, on which my family played a major part - this was an unconscious blessing. By 17 I was technically among the best at school in basketball, but my progressing weakness was unmistakably setting me apart from a physical standpoint. It was finally understood as something out of place, motivating a medical investigation.

At 19 the opthalmoparesis started, which made me compensate by slightly turning my head as needed.

At 26 (year 2008) I finally got my diagnosis. The process from entering medical investigation until the breakthrough lasted for 9 years.

Today, at 28, I often wish an electric wheelchair or some such when I have a long walk (~100 meters) or upward walk ahead, or I often have people give me a hand on lifting me from too low seats and to too high steps, or fetching something from the floor or carrying a heavy load, but I've been trying to find my own ways of doing these as to remain stimulated and delay the symptoms (I also avoid over-exercise). The outdoors hangouts with friends are vastly diminished - I try to keep myself socially active by changing to appropriate environments.

On a more psychological note, I see my condition as an ok thing. I have my ups and downs, but having gone through therapy for some years now, I've had a long way in understanding myself and the things that passed. I now realize most of my issues came not from my health condition, but from completely unrelated issues. This really helped putting Pompe into perspective.

Today, my goal is to be an active patient and make the advent of this condition a positive reason and motivator for my life, for every patient and the people around me. In time. I became vocal and advocating, took responsibility as a citizen and started to change my way among society.

This disease really changed me. I'm entirely different and wiser as a person. The weakness is a reality and I think the main lesson is learning how to see it in other terms. You're not like everyone else. This is neither good nor bad. You're just yourself. Traveling through life on your own terms, finding your path and uncovering the mysteries intrinsic to your own life is what I think means at the end. This is my philosophy.

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PS: Here goes a story that I shared with my pompe peeps in the mail list.

My symptoms really hit me on the late teens, so I went through the phase being scared of the future from a professional and financial point of view. I won't lie. I had a breakdown. It's really hard when you realize you'll still have to go through some chokepoints in life, like trying, if possible, financial independence or creating a family of your own.

In my worst days, I talked through tears to my father and said I feared the possibility of staying unable in bed in some future. He responded in the surprising, yet tender way he always do. "Do not fear your destiny", he said, "If you'll be bound to a bed or if you'll be cured, be happy for the life you'll have. Do not tie your happiness to things that are not on your hands. These are God's realm and the world's capricious designs. Do whatever you can to be happy, and rest easy on your work." I hugged him and it simply ended the subject for me. This was the answer I sought. That day I took my first step towards overcoming my condition. I've been on the road since.

Yes, he has this wise man atmosphere about him sometimes.