



Niemann-Pick Disease

Living Beyond My Rare Disease | Evren Ayik

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I live with acid sphingomyelinase deficiency (ASMD) or Niemann-Pick Disease Type B. ASMD is a rare disease caused by the deficiency or lack of an enzyme needed to break down certain fats in the body. I was three years-old when I was diagnosed, but I didn't know I had a disease until later in elementary school. My mother didn't talk a lot about it; she believed I should enjoy my childhood and live my best life.

Eventually I realized I was different from other children. I had compromised lungs, stiff joints and an enlarged liver and spleen, which kept me from participating in gym class and made sitting with my legs crossed and exercise difficult. Over time my peers began to take notice of the differences that the disease caused, and I was often excluded. Junior high and high school are challenging; having ASMD made it even harder.

But I didn't let ASMD stop me from enjoying life to the greatest extent. I found fun and adventure in Boy Scouts. I took road trips and enjoyed all the fun diner and restaurant stops with my grandparents. I went geocaching as often as I could. (I'm up to over 600 finds!) In elementary school, I started skateboarding – even though it was dangerous with my enlarged spleen. I was really careful! But the one thing that was too dangerous that

I *really* wanted to do? Paintballing! At first it was impossible because of the risk. Then my doctor suggested a baseball catcher's chest protector, which completely covered my spleen, and I was free to join my friends on the paintball field.

In high school, I joined the book club, an advocacy group for students with disabilities, and other youth groups. I began working as a peer-assisted learning aide for the school's special education teachers, as well as an office assistant. I was eventually recognized as being a positive role model and was awarded a Rotary Youth Leadership Award. I also earned the rank of Eagle Scout and was chosen as a California Boy's State representative. By graduation, I created many happy memories for myself because the thought of dwelling on my problems and wasting my opportunities to have fun never crossed my mind.

When I was 16, I was given the amazing opportunity to travel to the FDA in Maryland, where I spoke about the impacts of ASMD on my life, which are many. ASMD has affected my skin, eyesight, joints, liver, heart, hearing, spleen, blood, bones and lungs—quite a long list. After my first talk at the FDA, I was invited to speak in other forums to raise awareness about ASMD, including a corporation, an international webinar with other rare disease advocates, a conference, and awareness and charity events. My most recent advocacy project is a book for children with rare diseases, which I co-wrote with my mom. It is currently being illustrated, and my mom and I hope to publish it before the end of the year. It is a book that I would have liked to have had while growing up with a rare disease. My mom and I are also co-bloggers for the National Niemann-Pick Disease Foundation, which is the US organization representing ASMD and Niemann-Pick Type C families. In August, I was proud and thrilled to learn I had been selected to receive a prestigious TORCH award from Sanofi Genzyme in recognition of my leadership and advocacy.

When I talk about having ASMD, I don't just talk about how the disease affects my life. Although it's super important to help people realize that real, living human beings—babies, children and adults—live with diseases like ASMD, speaking only about my disease feels one-dimensional. I am a multi-faceted person with values, purpose, gifts and dreams. I do not believe that my only purpose in life is to advocate for people with ASMD, but I am proud and willing to share my story with others so they can meet and get to know a human being who happens to have a rare disease called ASMD.

If you are reading this right now, who are you? Are you seeking out opportunities for accessible fun and adventure as you simultaneously stand up for what really matters to you? Do you know who you are beyond your physical features? That's what it means to live beyond your disease. So go for it!