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Anthony

Born January 1993 | Bedminster, New Jersey

Living with DMD

Anthony maneuvers himself down the ramp behind his family’s suburban home, which is surrounded by sprawling acres of picturesque New England farmland. The sun is beginning to set, but it is unusually hot for an afternoon in May—too hot for Anthony to spend much time outside because sweating bothers him. It causes discomfort and itching, and drying off and changing is a lengthy ordeal he would rather avoid today. Still, there is something he wants to do. His family, led by his 14-year-old younger brother, Robbie, has a fascination with model airplanes. In an oversized garage, they maintain an entire workshop dedicated to the assemblage and repair of these planes, which require years to learn how to fly. When they were younger, the boys would share the task of piloting the planes, with Robbie serving as backup to Anthony’s set of remote controls. Together Anthony and Robbie would make the planes zip across the yard within feet of the ground, shoot vertically up in the air, and glide gracefully to catch the wind just before flipping upside down and nose diving with a quick recovery to avoid crashing. This hobby bonded the brothers together and gave Anthony a sense of freedom and movement at a time when his own body was deteriorating and his physical capabilities dwindling. These days, Anthony can no longer manipulate the remote control with his hands, but he still enjoys watching the planes, which Robbie jokingly flies directly toward Anthony, changing course at the very last second.

Anthony lives with Duchenne muscular dystrophy (DMD), a genetically inherited disorder which affects muscle function progressively

throughout one's life. Anthony showed signs of the disease as young as three years old. He presented with Gower's sign—relying heavily on his arms to stand up. "I remember the day we got the diagnosis" says his mom, Beverly. "I felt completely out of control." Anthony and his family began to grapple with the implications of living with a degenerative muscular condition. "Early on it was really hard for me to accept it," remembers Anthony. "I didn't know much about it. My parents would tell me about it, but only if I asked."

At eight years old, Anthony first had to face the long-term and irreversible effects of his condition. "I tripped over a video-game controller and landed on my femur." The fall resulted in a fracture to his largest bone. "That day is forever. I'll never forget that day." The excruciating pain left Anthony with few options but to start using a wheelchair, which his mother had already purchased, fortuitously. "We ordered the wheelchair before we really needed it," his mother recalls. More importantly, she had the house modified to suit the wheelchair

prior to his fall. "Once he really needed it, and was in the wheelchair, we knew we would have to redo the whole house."

As his physical limitations became more obvious to peers, Anthony began honing his ability to overcome the psychological burdens placed on a young man in his position. "My mom came to school and taught the kids about Duchenne," he remembers. Anthony too, began to speak up about the challenges he and other boys with Duchenne faced—the early premonitions

of a patient advocate in-the-making. "I came in and talked to my class and showed them a video about Duchenne. What I like about that is that it shows people that it's not something to be afraid of just because they didn't know what it is." High school was a challenging time for socializing, as Anthony's disabilities became more obvious to his peers. "At some point I decided to start talking about it. One of my English classes invited me in to speak and then I spoke to every English class at the school. So, I did about 20-30 talks. It really opened people's eyes, and it allowed me to open up too, because people understood me better."

After graduating from Rutgers in 2015, Anthony found it difficult to transition into the professional world, despite his impressively high marks in school. "The two years after college were a tough time for me," wrote Anthony on his Facebook page. "I wanted so badly to start my career and be a beneficial member of society." It seemed that no employer would look beyond his obvious physical limitations—Anthony uses a power wheelchair and a breathing machine—both results of his Duchenne muscular dystrophy (DMD). The neurodegenerative disease took away much of Anthony's physical abilities, but he refused to let it become a roadblock to sharing his intellect. Anthony started his own web-design company as a high school freshman and later studied Communications in college, graduating Summa Cum Laude. Despite his skills, Anthony still had to battle the stigma of being "disabled" in a professional sphere designed for the "able-bodied." "No one seemed to be willing to look past my disability and provide me the opportunity to prove myself."

One day a friend recommended that Anthony try to work in the field of pharmaceuticals—specifically companies that developed treatments for rare diseases. Since Anthony was young, he had been

interacting with members of the Duchenne community as well as the larger rare disease community, drug developers, and members of Congress. His friend thought that perhaps a company that prides itself on curing rare disease would be ready to see Anthony's experience as a valuable asset, not a hindrance. "I've always been big on patient advocacy," says Anthony. When a pharmaceutical company offered him an interview, Anthony was overcome with excitement, and apprehension.

Nerves aside, Anthony nailed the interview and got the job. However, he remained uneasy about what it would be like at the office each day; and how he would be perceived by co-workers, using a power wheelchair. "Some people see you in a chair and they think you have mental disabilities, or you're not smart," he explains. "And it's hard to get someone to look past that and see you for your abilities and what you have to offer." Fortunately, his office-mates were quick to embrace Anthony for who he was and saw the intrinsic value of his work. He recently has transitioned into

being the Master Team Integrator for the IT department, where he develops new tools for communication between the various working groups at the company. He only commutes to the office one day each week, but always looks forward to it. It provides relief from the stigma of "disability" and allows Anthony to show his productivity.

These attributes have served him well over the years, allowing him to make friends and progress through school despite the unavoidable setbacks and complications rendered by DMD, chief among them, widespread muscle weakness. "The heart, the lungs—really everything is supported by muscles. So you have to get used to everything weakening. When muscles supporting the lungs weaken, you can't breathe. When the heart weakens, your blood doesn't flow. So there are a lot of things you have to do to offset the weakness." Anthony's fortitude and willingness to tackle these immense physical deficits has undoubtedly brought him to where he is today.



At the height of his university studies, Anthony contracted pneumonia, forcing him from class for weeks and stifling his ability to breathe. The bout with illness convinced him to put down his misgivings and try a ventilator—the only viable option to get the oxygen he needed. “After going through that and seeing how difficult it was for me to breathe, and all the struggles with pneumonia, I thought, *Why not do something to prevent that?*” His prior reticence to try the ventilator was due, in large part, to the

social stigmas that he felt came with it. “I was very self-conscious about being seen in public using it,” recalls Anthony. He had resisted the device for years despite his doctor’s recurring recommendation, but finally accepted the therapeutic necessity. “There comes a point where being stubborn is not beneficial to your health.”

Adapting to the ventilator was difficult for Anthony. He had been breathing for himself his entire life and undoing a lifetime of

muscle memory was not simple. It required practice—like learning to write with the non-dominant hand. “I would feel like I wasn’t getting enough air,” recalls Anthony. “I was trying to breathe for myself.” He was challenged by the prospect of letting the machine breathe for him. He was afraid to sleep. “One of the big things was that I started by using a full-face mask, so I couldn’t call out for help when I needed to.” This inability to communicate was unnerving for Anthony and his family, but he remained



determined to work it out. Getting a nurse alleviated much of his anxiety, knowing that someone was there to monitor and help him. "Before the nurse, we were hesitant having him wear the mask," recalls Beverly, his mother. "We couldn't hear him through the monitor. He couldn't really talk. If he got sick or threw up, we knew he could suffocate himself. Once he adjusted Anthony could go to bed without fear. He continues to utilize his respirator and mask today, taking breaths every few moments as he speaks.

As part of a population of young men aging with DMD, Anthony's lung health is central to his longevity. Mechanical ventilation has helped dramatically change the prognosis for DMD, which once stood below 20 years of age when many young men died from respiratory failure (the other main cause of death being cardiac failure). The extended life-expectancy today is 25, which is in part due to increased respiratory therapy, specifically the ability for families to use the equipment regularly at home. With this in

mind, Anthony overcame his reservations. He uses his ventilator daily.

"I also use an Ambu Bag," he explains, pointing out the device. "It gets placed over my face and then squeezed manually. I use it for air-stacking—stacking one breath on top of another, which helps to increase the volume of my lungs." Anthony's doctors have seen an improvement in his lung capacity over the last two years since he became committed to these exercises.

These medical interventions are prolonging Anthony's adulthood and allowing him to pursue some of his dreams. In addition to his job, in which he takes great pride, he has developed interests in video games and computers. "I got into web design," he says. "I have a web-design company where I create websites for organizations. This got me into communications and marketing and doing the work that I do now." He also has burgeoning interests in writing music, both the instrumentals and the lyrics. He creates

the music digitally and then hands the material over to a local band, who performs and records it. His songs are well-regarded in his community of friends and family and he has shared them as background music to digital photo montages of his friends at a Duchenne summer camp, which he attends annually.

With a solid foundation of family support, personal assistance, and a fervent charm, Anthony continues to contribute valuable

insights to the field of rare disease. "Be grateful for what you have," he professes. "I'm definitely grateful for everything I have. There are a lot of things I can't do, but there are so many things that I can."

