LIMITLESS

Stories of Defiance, Opportunity, and Ability

Preface

What you hold in your hands is a collection of dreams and aspirations that refuse to succumb to the challenges of living with a rare disease. Although these conditions are often referred to as "life-limiting," *Limitless* tells the true stories of nine individuals living life to the fullest, decisively showing us how their abilities and talents have flourished despite battles for their health and equal opportunities. Each of these extraordinary people and their supportive families, have pushed their limits—faster, higher, and further beyond the expectations placed on them by society.

Limitless is an emblem to the remarkable ingenuity, creativity, and resilience of finding one's place in life, against the backdrop of a harrowing prognosis. It also holds a mirror up to society, reflecting how it treats those who wear the assigned identity of "disabled" as they pursue the lives they both want and deserve. At times there is frustration with the indifference toward them, with the lack of attention paid to their needs, and with the stigmas that (at times) relegate them to being "less-than" or "other." In addition to the battle for their own health, many of these individuals expend immense energy fighting against these pervasive stereotypes, to the point where many of their greatest successes stem from overcoming prejudices and gaining social acceptance. Although this is commendable, we join our friends in calling upon a more hospitable and gracious society, where abilism is replaced by a greater sense of appreciation and a dignification of difference.

At Living in the Light, we strive to illuminate the unseen dimensions of a person's life that too often get covered beneath the weight of living with a rare disease. In doing so, we deepen our shared understanding and vision of what it means to be a human being faced with the unknown. As listeners and writers, we uphold the redeeming quality of a person who is truly being heard. As viewers and photographers, we regard seeing as a conscious act with the ability to empower and enliven those seen by the way we look at them—with reverence and compassion. From this combined perspective we learn a lot and feel there is a profound need to listen to these stories and make more space for the voices of these storytellers to be heard.

It is our sincere hope that *Limitless* encourages each of us to consider the role we can play in lessening the limits we place on each other. Together, we can make greater efforts to improve the accessibility and design of our homes, schools, and workplaces. We can improve professional opportunities, as well as our transportation systems, our institutions, our cities, our countries and most importantly our hearts, to be more inclusive and hospitable to all. This project comes as a tribute to those who continue to seek this change, be it through groundbreaking science, heartfelt patient advocacy, or a deepening of the ability to listen to our fellow humans a little better.

And so, with your attention, we invite you into the houses, backyards, dorm rooms, and onto the courts and trails that these people call home. We hope you can read each story with an eye for detail, critical thinking, and most of all, an open heart. It is this type of emotional intelligence, garnered from close reading and viewing, that has an untold capacity to heal those around us.

Levi Gershkowitz
Founder and Chief Executive Officer
Living in the Light

September 2018

THANK YOU

At Santhera, we have a set of core values that guide our decisions and actions every day: Commitment, Passion, Respect, Collaboration, and Accountability. And while we strive to infuse those values into all our efforts, nothing illustrates them greater than these stories that have so graciously been shared with us. We can't thank the families portrayed here enough for opening up your homes and lives to the Living in the Light team and for allowing us an intimate view of your challenges, successes, hopes, and dreams.

Santhera aims to develop treatments for rare neuromuscular and neuro-ophthalmological diseases. Our commitment comes with a unique sense of importance and urgency: to save eyesight; to stabilize breathing; to preserve motor function; to maintain and improve the quality of life of people living with rare diseases, and by extension, their families and communities. The loss of one of our beloved participants, Kyle, during the creation of this project is a stark reminder of that urgency. For our patients, each day is fragile and not a single day of health is guaranteed. The need for therapies remains great—there is still much work to be done. We dedicate this book to Kyle's family, who continue to carry on his memory.

To all the families out there, we will continue to work hard for you, to share your stories, and help raise awareness of each condition. Please accept this book as a token of our appreciation for your involvement in this project and for sharing your experiences with so many. Your stories provide the compass for our work.

With appreciation on behalf of the Santhera team,

Jodi Wolff
Head of Patient Advocacy, U.S.
Santhera Pharmaceuticals (USA) Inc.

Contents

Amin

Born June 2006 | Los Angeles, CA

Living with DMD

There is something striking about Amin's eyes, as if they are two deep jade pools of water that pull you into the quiet wisdom that streams from them. At 11 years old, his presence is poised, elegant, and stoic; until you reach his toy car collection and he lights up like the bright child he is, talking about Lamborghinis, Bugattis, and the fastest vehicles in the world. Amin's parents, Mastooreh and Ali, open their home with gestures of hospitality—offering house slippers to their guests so they can take off their street shoes. The family emigrated from Iran in 2015, bringing the beauty of their Persian culture with them. A perfectly cooked three-course meal sits waiting at the dining table, with cups of Persian tea, and treats for dessert to follow. Instead of cooking a traditional Persian feast of *khoresh* (stew), Mastooreh baked lasagna and a cheesecake to better cater to American cuisine. She still included a traditional Persian side-dish of saffron-rice with cranberries, and *tadik* (a crunchy rice delicacy).

Ali, Mastooreh, and Amin live in an apartment in Beverly Hills with panoramic windows that let in the light and vistas of the surrounding hills and city life below. While sharing their delicious meal, Mastooreh and Ali gracefully describe their most difficult journey—their son's Duchenne muscular dystrophy (DMD) diagnosis and their emigration to the United States, to find the best available treatment.

Back in Iran, Amin was a happy child, growing up in relative comfort. Ali was an experience physician who specialized in emergency medicine, while Mastooreh, a painter and illustrator, taught art at the local university. Coming from prominent families, they had more-than-sufficient income. They lived in a beautiful home in Tehran, surrounded by Amin's grandparents and a large extended family.

"Iran is very rich in natural resources; oil, gas, and gold. However, the most beautiful resource in our country is the people. There, we supported and depended on family."

When Amin was five years old, his kindergarten teacher expressed concern about Amin's physical limitation because he couldn't run as fast as his classmates. "She asked us to visit the orthopedic doctor," remembers Ali. "We did some blood tests that showed Amin had problems in his muscles. The doctors sent his blood samples to Germany and it confirmed that Amin had [Duchenne] muscular dystrophy."

Mastooreh found a DMD organization in Iran that held meetings twice a year. Fortuitously, a meeting was scheduled the same week they received Amin's diagnosis. The family attended the meeting together, and Amin met older boys with DMD who used wheelchairs. At five years old, Amin could still run and walk. "In that moment," Mastooreh remembers, "Amin figured it out. He understood the reason why we were there next to all these people who have Duchenne." Amin said he adapted quickly to the realization. "He was always very accepting," says Mastooreh with a tear in her eye. She continued to bring Amin with her to the meetings so he could see the boys older than him with power wheelchairs.

Amin participated in physical therapy and was swimming nearly every day, but in Iran he did not have access to treatments or clinical trials. A doctor, Ali had access to information about emerging therapies and was determined to find the best for his son. "I always follow new clinical trials in the US and I knew one was starting soon."He decided to attempt the long journey with his family to travel to the US. "This medicine is important for all the world," says Ali. "And it is only in the US and Europe."

Obtaining green cards for the whole family was a huge hurdle. They hired a lawyer to help them. "The only way our lawyer could get our green cards was to make an investment for an EB-5 Immigrant Investor Program, with a payment of $500,000." After three years they were still waiting in suspense for a response—with no green cards in hand. "We were concerned about missing the trial and we couldn't wait for the green cards' long processing time. Amin was losing his ability to go up and down stairs. We decided to come to the US with a medical visa."

The family traveled to Dubai to request a medical visa from the US Embassy. After a year waiting, Ali was finally granted a visa but Mastooreh and Amin were not. Ali emailed several embassies throughout the world to request help, but resigned himself to the fact that he might be leaving for America alone. Just a few days before

"In our country we were dependent on a huge family. But we couldn't tell them. They just knew we left; they didn't know why. We could not believe or imagine we would be moving to America." *–Ali*

Ali's scheduled departure Mastooreh and Amin miraculously got their visas and Ali rescheduled his flight to leave a week later, this time, with his wife and son. Without telling family and friends besides their own parents, they rapidly packed a few suitcases and left Iran. It was a gamble. Mastooreh and Ali didn't know for sure if they would be leaving permanently or what exactly they would find. They arrived in the US in March 2015, and a year later, in March 2016, they received their green cards.

The family received medical visas based on Amin's appointment with John Hopkins University in Baltimore. However, Ali's cousin recommended they visit UCLA and then decide their next plans. At UCLA, they met experts researching DMD.

"I believe the US has the best medicine and safest trials in the world," says Ali. "There are seven billion people in the world looking to the US for new medicine. Everything is here for patients: new medicine and new clinical trials." In Iran, university budgets do not have much funding available for research, studies, treatment, or new science. "This is hard for people who have to live and can't move to another country," says Ali. "We decided to move to a country that we never saw; that was huge; and far, far away. We took a risk!" exclaims Mastooreh.

Amin started fourth grade in the United States. "When I started school I didn't know any English," he recalls with a smile. His father remarks, "After almost three years, he's better speaking than us!" Mastooreh chimes in. "Amin's school homework is homework for all of us too." They all laugh. Then, the conversation turns to a more somber tone, as they talk about Amin taking his last steps. He was nine years old, and the family was out at the UCLA pool for physiotherapy. When the time had come to leave, Amin was waiting by the car and suddenly lost his balance. He fell, breaking his right femur in two places. The fractures required surgery, which was long, difficult, and very stressful for the family. "When we were in the emergency room, I was crying, crying," recalls Mastooreh. "Amin said, 'No mom, don't cry. God is very strong. Trust to God. Trust to God.' He makes *us* strong."

"When we were in the emergency room, I was crying, crying. Amin said, 'No mom, don't cry. God is very strong. Trust to God. Trust to God.' He makes *us* strong." *–Mastoofeh*

Since that incident, Amin has used a wheelchair, which he remains positive about. He especially enjoys his ability to maneuver the wheelchair at fast speeds. This developed into a love for sports cars and anything that moves fast as well as power soccer, an adaptation of soccer for power wheelchair users. He recently joined a power soccer team and it is the highlight of his week. At Sunday practice in the gymnasium, the basketball court is split in two halves for the junior practice and the varsity practice. Amin started out on the junior team, practicing passes and general ball handling. At the far side of the court, the varsity team moves with great speed and agility in full scrimmage, with noises rising of competitive sportsmanship and the occasional clang of a collision. Amin moves calmly, swiftly and precisely, a glimmer of excitement shining in his eyes. From the back of his power chair hangs a piece of paper, "AMIN #2, LA AVENGERS" with a red Bugatti sticker he has pasted below.

Like the newest cars, Amin and his parents look toward groundbreaking biomedical technology to hopefully treat DMD in his lifetime. It is a source of fascination for all of them, who remain determined to help Amin get the best treatments available. This willpower has led them on an incredible journey, one that has taken him halfway around the world in the search for a cure.

"My main focus now is to create more adaptive, accessible, universal, and barrier-free designs. And there's a very, very, very small minority of the architectural world that addresses that."

Jason

Born April 1985 | Columbia, MD

Living with DMD

Jason and Adele were tired from their day of travel and arrived at the Berlin central train station late in the evening, ready to get some rest. The mother-son duo had been in Prague for nearly two weeks, where Jason was studying sustainable architecture, with his classmates from the University of Maryland's School of Architecture, Planning, and Preservation. "Our class was combined with a bunch of students from Prague," remembers Jason. "We learned about designing buildings that were more efficient—that use less fossil fuels." Jason was exhausted from the long train ride, and needed to find a bathroom before taking a cab with Adele to their hotel.

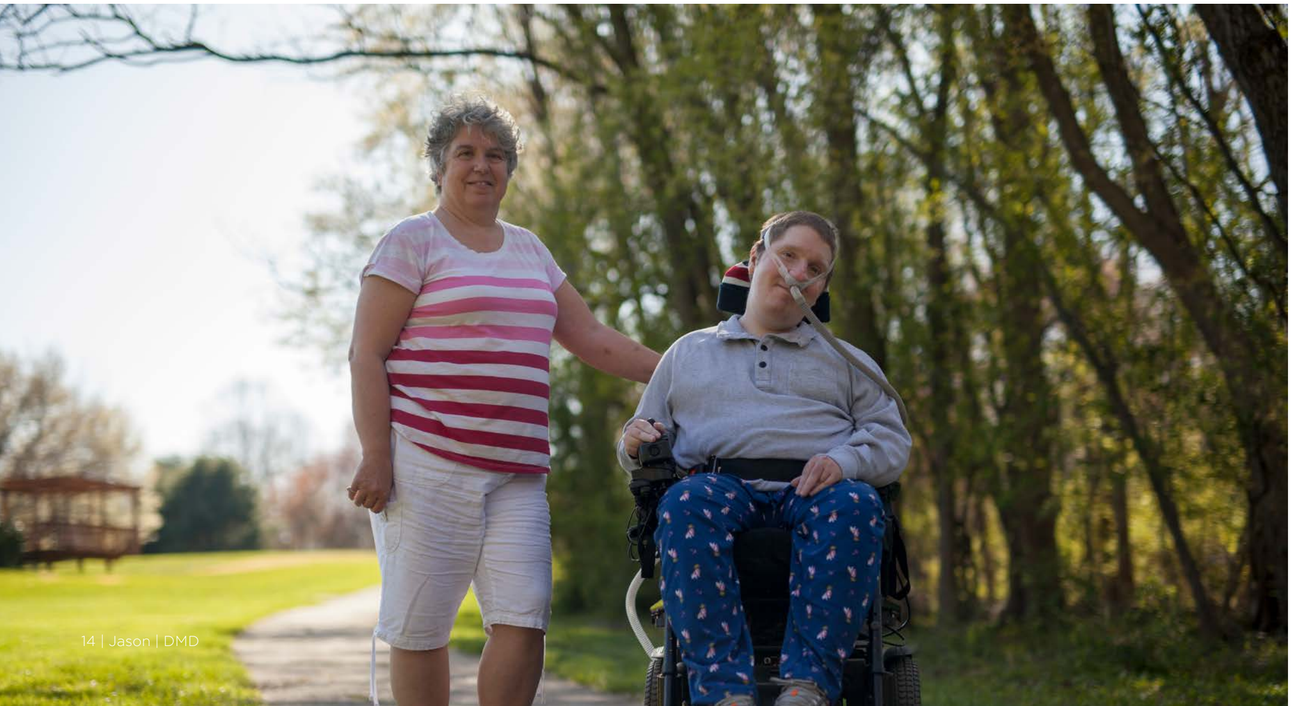
The attendant led them over to the "accessible toilet." Jason rarely held high expectations about such devices, especially in public train stations. He was used to struggling to get on and off the toilet and back into his power wheelchair. Even in bathrooms with multiple grab-bars, his muscles were often too weak to lift himself onto a normal toilet. Jason was born with Duchenne muscular dystrophy (DMD), a neurodegenerative muscular condition that irreversibly weakens one's muscle strength over time.

However, as a wheelchair user, Prague was a welcome surprise, especially the public transit system. "About 90% of the public transportation was accessible," says Adele. The two were delighted to navigate the medieval city with relative ease. A similar surprise was in store at the train station in Berlin. As soon as the attendant unlocked the door and swung it open, Jason and Adele were taken aback by what lay inside. "It had to be a $40,000 toilet," she exclaims, describing "the king of toilets." "Everything was on a track. The toilet went up and you could adjust the height of everything... It was in a damn train station!"

True accessibility had been rare in Jason's life, especially living in the US, where accessible designs are often neither thorough nor valued. "In Prague they don't have the ADA [Americans with Disabilities Act] like we have, but they have common sense," says Adele. "Over here, when they want to make something accessible, they do it cheaply. Over there, if they want to make something accessible, they do it so that it works for the person."

The pursuit of barrier-free and universal design has become both a necessity and a passion for Jason. A wheelchair user since the age of 11, he lives in a world where solutions to his needs around mobility are rarely well-developed. Harnessing the ability to redesign buildings and devices is a tangible route toward empowerment for the aspiring draft architect—transforming hardship into constructive acts. Yet, within the field, Jason has continually struggled against physical and social barriers. "Just because I'm in a wheelchair, I've had to spend a lot of time proving my cognitive abilities," says Jason. "I think that's one of the major misconceptions. Especially now that a lot of people are graduating from college." Against societal preconceptions, he continues to fight for his dream of an influential career in architecture.

Back at his home, in Columbia, Maryland, Jason's eyes rest firmly on his laptop screen as he works hard to master a new virtual drafting program. "Most design happens digitally now," he explains. "But when I was applying for school and jobs, I would be asked to provide hand-drawn sketches... I obviously couldn't do that." Getting accepted to the University of Maryland was no small feat for Jason, but it paled in comparison to this past year when he graduated. "Finishing my bachelor's degree was life-changing," says Jason. "I feel like it was a true test for me, and I passed."



To celebrate "passing the test," Jason and Adele are busy planning his graduation party, which coincides with his 33rd birthday. Jason was first diagnosed when he was four, in 1988. "At that time there was not much known about Duchenne other than the name of the disease, that it usually affects boys, and that they're usually dead before they reach 15 or 16," recalls Adele. In 1989, geneticists discovered the gene sequence that caused DMD, and in the decades since, research has grown phenomenally, leading to advances in management and treatment, but as of yet, no curative measures. Whether they find a cure or not, Jason dreams of helping create a world better suited to his and other people with disabilities' needs.

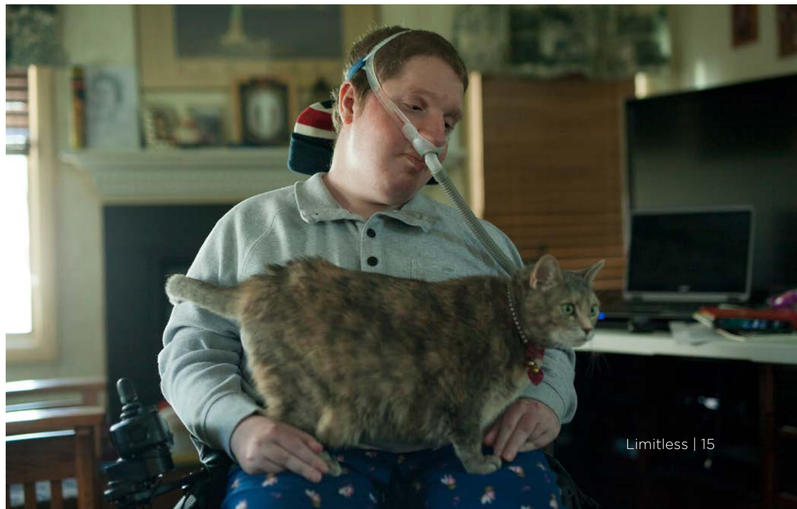
The inspiration to apply to an architecture program came from Jason's own experiences with accessibility issues as well as his innate desire to build. "When I was a kid, I always wanted to do something in building, because my favorite toy was always Legos," says Jason. "I think that's where it all started from." As Jason's needs progressed, his home became a venue for trying new layouts. To make Jason's abode more comfortable and utilitarian, Adele and an uncle did much of the renovations themselves, which helped inspire Jason to try out the process of designing a space.

For his college entrance essay to UMD, Jason studied the life and work of Charles A. Cofield, a quadriplegic architect, who graduated from MIT and UCLA, and worked for the Los Angeles Housing Authority, bringing better housing options to low-income areas of the metropolis. Studying and learning to identify the intersections of ableism, urban planning, and design helped spawn Jason's interest. He also became enthralled with the work of the influential architect, Michael Graves, who started using a wheelchair in his last decade of life—leading to a flurry of new architecture—before passing away in 2015.

"There aren't many disabled architects," says Jason, discussing the lack of accessible design being done by individuals who rely on them. From his disposition, Jason sees the opportunity to offer something to the field that few others can: chiefly, first-hand knowledge of which barrier-free and universal designs work the best for people in power wheelchairs. "A lot of architecture firms deal with healthcare buildings," he explains, noting the extensive experience he's had navigating hospitals. "I think I would be able offer that industry something that it usually doesn't have. They don't have anyone who works and designs from my point of view." Using virtual environments, Jason is now able to design entire structures, devices, and urban spaces that can be fully explored on a computer before physical models are built. Mastering new technologies is crucial for Jason to meet his goals. "My main focus now is to create more adaptive, accessible, universal, and barrier-free designs. And there's a very, very, very small minority of the architectural world that addresses that."

Even in some of the most progressive cities, like Prague and Berlin, Jason still sees room for improvement. "It's hard to find a [hotel] room that has enough power outlets to charge my equipment," he says. Jason uses a non-invasive respirator around the clock, combined with his power wheelchair, a powered air-mattress that he sleeps on, and a whole host of specialty machines to support his health on a daily basis. "At the hotel in Berlin, they gave us a king-sized bed," he says, chuckling from behind his respirator. "I guess they thought we could've been a couple."

"Sometimes we're like a mother and son, and sometimes we're like an old married couple," Adele quips in her Bronx-born accent. Her loquacious and fervent demeanor has served the family well over the years, as she grappled with the implications of DMD, sought out resources and treatment, and supported Jason in his day-to-day needs—almost entirely as a single mother. "We spend a little too much



time together," admits Jason. To support the family financially, Adele became Jason's nurse through Maryland's Developmental Disabilities Administration, getting paid for her son's demanding at-home care-taking. "He was the cutest baby I ever saw!" Adele reminisces, as Jason rolls his eyes. She goes on to tell us how Jason and his older brother, Josh, were often confused for girls when they were toddlers, because they had such long eyelashes. Jason and Adele's sardonic chemistry is unmistakable and there is no lack of jeering between the two, although underneath, it is clear that there is a solidarity and love that few could fathom.

Within the Duchenne community, the mother-son duo have found themselves a sort of model of their own—bringing important information to newly diagnosed families, and telling their story. "It knows no socio-economic boundaries," says Adele. "It hits rich and poor—many of the families who have Duchenne are low-income, and they don't have an excess of time to investigate on their own." She and Jason are elated to help answer many of the questions that were simply unknowns when he was first diagnosed. "I've spoken at Parent Project Muscular Dystrophy," says Jason. "I've done a lot of speaking. My neurologist speaks to a lot of college graduate students at Hopkins. Every so often, she invites me to speak with them."

As Jason remains focused on his drafting prowess, hopefully leading to a career, he continues to battle the physical effects of DMD. In 2001, he underwent spinal-fusion surgery to correct his severe scoliosis, which was affecting his lungs, and now has transitioned to using a non-invasive respirator full-time. However, his overall health is rather uncanny, as many younger men he has befriended in the community have passed away in their youth. "It's tough to deal with." Jason ruminates about losing friends in the DMD community. "It reminds me of my own mortality, with maybe some feelings of guilt mixed in: How come my health is still decent? In the same breath, friends dying helps me stay motivated and keeps me moving forward. There's a lot on my plate that I want to do in however much time I have left. I wanna make that time special."

Kyle

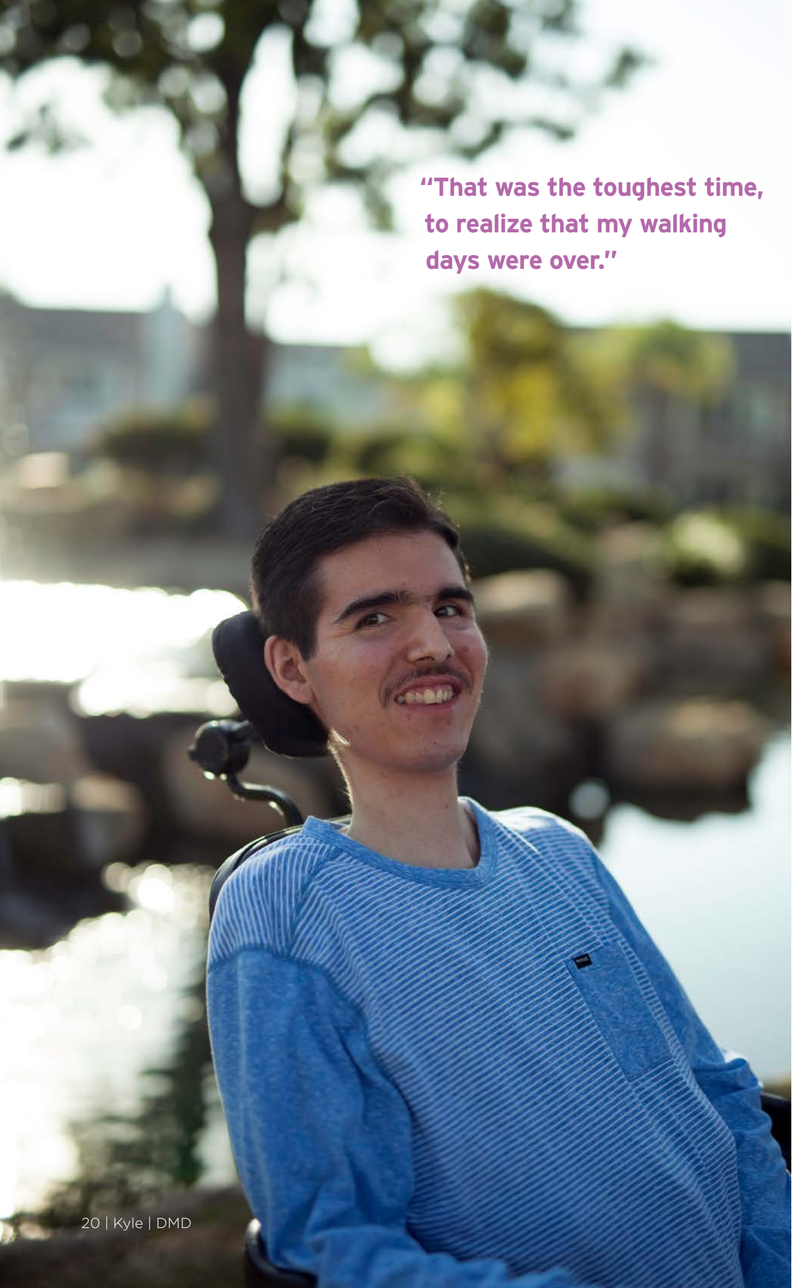
January 1995 - July 2018 | Yorba Linda, CA

A Life with DMD

The oversized soccer ball shoots across the polished wooden floor as the wheelchair athletes practice their passing game. Hands on his hips, their head coach, Armando, explains different strategies and positions from which to send and receive a pass—the architecture of a sport called power soccer. The players listen closely from their power wheelchairs. Among them there is a vast range of age and life experience, from boys and girls barely older than seven, to adults in their 30s and 40s. They coalesce each Sunday in a gymnasium in Glendale, California, to practice the sport they love.

Kyle quickly maneuvers his power chair into position to demonstrate the different ways to pass. His experience is a gift to the team as an eight-year veteran of the sport and the head coach's son. With striking agility he makes precise moves with his chair and volleys the ball to the other players, giving them an opportunity to understand the necessary reaction time, the chairs' turning radii, and the correct speed—all instigated by the deft moves of one hand on the chair's pivoting joystick. With each pass, the joy of the sport emanates from Kyle in his furtive smile.

"In my heart, I have this sense that I want to make a difference in the world—not just for me—but for others with Duchenne."



"That was the toughest time, to realize that my walking days were over."

"About six years ago, they started making a wheelchair strictly for power soccer," Kyle explains. He displays his chair, called the Strike Force, and its swift capabilities. Just a minute gesture with the control stick unleashes a rapid, aggressive movement from the chair, allowing for powerful "kicks" by utilizing a welded cage around his footrest. "It's lower to the ground. It has more torque. You can turn quicker. It makes the game more fast-paced." The Strike Force chair is an emblem of how far this sport has come, from grassroots beginnings on vacant basketball courts in France and Canada pre-1980, to an internationally recognized sport with its own tournaments and leagues under the banner of FIPFA (Fédération Internationale de Power Football Association).

The progression of the sport has mirrored the advancements for people who use wheelchairs around the world, providing an outlet for competition and teamwork. It has helped defy the falsely-portrayed archetype of the sedentary wheelchair user, of the "disabled" person lacking any recreational pastimes. For Kyle, who was diagnosed with Duchenne muscular dystrophy (DMD), a neurodegenerative muscle disorder, at age six, the sport is far more than a passing hobby or weekend diversion—it is a pivotal event he looks forward to each week with pride and aspiration.

"It's been pretty life-changing for me," he relates. "To have an activity to do—an outlet." Kyle was born into a family ensconced in the joy of sport. "He's always liked sports," says Armando, who himself fosters an enthusiasm for athletics. "When he was five years old, he played basketball and t-ball." During games and practices, Kyle struggled to match his teammates physically. "I noticed when the coach would have him run around the bases, he was the slowest one. When they would run twice around the bases, the other kids would lap him." His parents brought their concerns to his pediatrician. "He had really large calves," remembers his mother, Sandra. "With Duchenne, your muscles are enlarged and then slowly waste away. I have the same problem, so I thought, that's just what I have. Turns out I'm a carrier."

Months of genetic testing revealed that Kyle had been born with DMD and he and his family began to cope with the facts of the terminal condition, which still has no cure. They learned Kyle's life expectancy was severely truncated (prognosis was late teens to early twenties, although it's extended by almost a decade now.) Dishearteningly, Kyle accepted that he would no longer be able to play sports, or so he thought. As the disease progressed, his muscles shrank, making daily activities more challenging. On his first day of school in seventh grade, Kyle felt unusually weak, and during the long walk from the parking lot to his middle school in Yorba Linda, he fell and couldn't get up. "One day I was walking and it was fine," he says. "Then, I just felt so weak. It happened so quickly." Sandra rushed over to help him up and make sure he was okay. Kyle would rise to his feet again that day, but with the knowledge that those days would be numbered. "That was the toughest time, to realize that my walking days were over."

Upon entering eighth grade, Kyle started using a power wheelchair, and he has not walked since. His friends and teachers stepped into his life to help facilitate new activities and show him love. "I was very lucky in that regard," reflects Kyle. "I had

friends that really looked out for me and helped me out. Some of the best friends that anybody could ask for." With a push from a supportive teacher, Kyle joined the yearbook staff in eighth grade, and stuck with it until he graduated high school. "I was assistant editor at yearbook for a couple years. Those were some fun times."

Armando and Sandra continued to support Kyle with his day-to-day needs, as well as finding him new activities. Taking him to watch sports became a family bonding experience. Armando consistently went out of his way to score tickets to see Kyle's favorite teams. They would go to Lakers' games in LA, Angels' games in Anaheim, and would even travel to see University of Arizona (Armando's alma mater) football and basketball games. Kyle's sister, Rebecca, was a soccer player and the whole family would travel to her games and tournaments. "I was her biggest fan," says Kyle endearingly. "But I was on the sidelines. I wanted to be independent and in control of the game."

So when Armando and Kyle first read about power soccer and a team based in Glendale, they didn't hesitate to just show up. Upon arriving, Kyle became elated with the challenge of learning a new sport and the potential for bonding with other individuals in wheelchairs. Likewise, Armando saw an opportunity to support his son's aspirations. "The first season, I just helped out as much as possible," he says. When Kyle's coach decided to move to Arizona, Armando saw a ripe opportunity to support his son and others who faced similar hurdles in life. After only two seasons, Armando stepped up to fill the vacancy as head coach, a role he has embraced for the past seven years.

Under Armando's guidance the program has expanded, from including mainly college-aged boys, to now including boys, girls, men, and women of all ages. "We have now grown to three teams." Each team is formed on the basis of age and experience. Armando continues to work at an insurance agency and coach on the weekends. "I work during the week. This is just something for Kyle and other kids who are out there. If I see a smile on their face, that makes me just want to continue doing it. It gives them something to look forward to—to be part of a team. Now they enter these tournaments, with other teams from different parts of the States. It's just awesome. It allows Kyle to meet people and play a sport they just love to play."

"A lot of these kids don't have much of an outlet," adds Sandra, who helps out when she can. "They are a bit isolated, socially. When they come out to play power soccer, it's a whole new world for them. They realize there are people like them." Kyle has formed numerous friendships through his team, which has provided a venue for socially active adults living with mobility issues.

The team has proved to be a more successful social outlet than higher education. Kyle attended a local university to study Journalism for a year-and-a-half, but felt socially isolated. "I decided it just really wasn't for me," he says. Likewise,

with the progression of Duchenne, daily activities had become more laborious. Even the act of eating became challenging, as Kyle's jaw muscles grew too weak to chew his food, resulting in malnutrition. "I have a g-tube now," explains Kyle. "To make sure I am getting enough calories. I was a little blindsided by that, even though I knew others with Duchenne who have the same problem." This recent development added to the list of reasons why staying at home was the best option.

"He would prefer to have his own apartment," says Sandra. "But I think he handles it pretty well." At home, Sandra and Armando prioritize care for Kyle, doing all the daily monitoring and therapeutic measures to help him retain mobility and avoid complications. Kyle understands the necessity of living at home, although he also strives for independence. One helpful measure is employment. Kyle works for his parents at their insurance agency in town, doing IT support for the franchise. "It's pretty simple stuff," relates Kyle. "Simple to you," Armando interjects. Clearly his work is valued and getting out of the house throughout the week helps Kyle avoid feelings of isolation.

Utilizing his prowess on computers, Kyle recently started his own fundraising and awareness campaign, upon turning 23, to find a cure for Duchenne. "I want to raise $23,000 this year," he says. "I know it's a big goal, but in the first month I've raised $3,000. It's a year long fundraiser, so I have time. In my heart, I have this sense that I want to make a difference in the world—not just for me—but for others with Duchenne."

Through Kyle's burgeoning social networks, as well as power soccer, he is learning valuable lessons from the broader community as he enters adulthood. "The main thing is knowing and seeing how people deal with different disabilities," says Kyle. "How they handle everyday life. How they are able to adjust and cope with it. They are such inspirational people. You can pretty much just do anything you put your mind to. It gives me a ton of confidence."

Back on the court, Kyle's poise is notable. His favorite position is wing, opportunistically trying to sneak around defenders for a clear shot at the goal. His aggressive, winding course forces the defense to re-adjust, scooting backwards at high speeds. Kyle darts between two defenders and sets up with his back to the goal—the standard pre-kick position. He fakes to the left, pulling a defender out of his potential shooting path. Suddenly, a pass comes from the center and in a blink-of-an-eye Kyle whips his chair around sending the ball careening toward the goal.

"I had friends that really looked out for me and helped me out. Some of the best friends that anybody could ask for."

On July 3, 2018, while traveling home from a national conference cup in Indiana where he had been competing with his fellow Vaqueros, Kyle went into cardiac arrest and had to be airlifted to the hospital. He was placed in intensive care for several days in Colorado, where he then passed in the evening of July 8th. Kyle was a courageous and fierce athlete, with an unwavering goal—to "knockout Duchenne" and improve his quality of life, while helping others. Among many of his virtues, Kyle is remembered for his joy and generosity. He continues to live on through his many friends, family members and teammates, as well as through his fundraising for research and the donation of his liver and kidneys. We are grateful for his immeasurable contributions and for being a trailblazer, on and off the court.

"Be grateful for what you have. 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."

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."

"I want to be as independent as possible, while I still can."

Tayjus

Born October 1995 | Cambridge, MA

Living with DMD

On a freezing day in Cambridge, Massachusetts, Tayjus drives his wheelchair up the ramp of Quincy House at Harvard University. The wide, heavy doors open automatically for him, but they haven't always been so welcoming. For two years, he advocated for a scanner that would read his student ID and open the doors so he could enter his own dormitory. His request was simple: "I'm a student in a wheelchair and I don't have use of my arms. I need a way to enter this building." After years of countless emails and phone calls, the university finally installed the requested scanner. Tayjus has been opening doors for himself and others ever since.

Tayjus lives with Duchenne muscular dystrophy (DMD), a genetically inherited disorder characterized by the progressive degeneration of muscles. Boys who are diagnosed with DMD appear to be born with normal muscular function, but soon lose their strength, beginning in their legs. Tayjus was diagnosed when he was five. He's unable to recall the early days when his parents say he fumbled and tripped while other kids ran, but vividly remembers the moment he got a wheelchair. "I was 11, in sixth grade, and it was getting harder to walk. I was very resistant to a wheelchair. One day when I stepped on a shoe, my ankle twisted and my tibia snapped." The doctors gave him a manual wheelchair, but he insisted on walking. At 14, he broke a shoulder bone when he tried to walk a short distance and fell. It became dangerous for him to walk at that point and he started using a power wheelchair.

A power wheelchair gave Tayjus more independence. He zipped through crowded hallways and lunchrooms, navigating school like any other kid. "In high school, I didn't think about my disability that much. I had a good group of friends and we had been friends forever. We were the academic types. I didn't have to give up things like sports," he says laughing, "because we mainly just played video games."

As he excelled through high school, Tayjus dreamed of attending an Ivy League university, where his bright mind and curious nature could continue to flourish. His college entrance essay focused specifically on DMD, and how his disability imbued him with a passion for social justice and a commitment to advocacy. His acceptance at Harvard was a moment of immense pride for Tayjus and his family. He was also nervous. What would it be like to have a stranger dress him and feed him? Would he be able to navigate the streets of a metropolitan city? Harvard

"People were nice, but interactions were forced, not meaningful. If I made a friend in another dorm, I usually couldn't visit them because their dorm was not wheelchair-friendly. If people said, 'Let's go out to lunch!' and I couldn't go, I'd come back to my room. I was trapped in my dorm. Alone."

seemed a universe away from the quiet, tree-lined New Jersey suburb where he grew up.

"My dad found an article about two girls with muscular dystrophy at Harvard. I met one of the girls and thought, 'If they can do it, then I can do it too.'" For a month-and-a-half of Tayjus' first semester, his mom stayed with him until he was settled into a new routine. Then one day he told her, "I think I've got this." With a sense of pride greater than her concern, his mom returned home, leaving her son, for the first time, truly on his own.

Tayjus' sparsely decorated dorm room, with its exposed concrete block walls and minimal windows, is contrasted by the lively and colorful way he talks; his conversational style is punctuated by expressive hand gestures and smiles that reveal his boyish dimples. He has control of the muscles in his hands as well as his head, neck, and face. His PCAs (personal care assistants) live in a room attached to his living space. They use a lift to assist him with transfers from his bed to his wheelchair; they provide physical support with eating, getting dressed, and using the bathroom. The PCA schedule, scribbled on a dry-erase board, reads as if Tayjus runs a small business, which in a sense, he does: He hires and trains his employees, manages conflicts, juggles complex schedules, and approves timesheets. In general, PCAs do not accompany him to classes or social outings. "It changes the dynamic when there is another person around. I want to be as independent as possible, while I still can."

The Harvard campus proved to be a huge challenge. "It's really backwards here. Only two out of twelve freshman dorms are accessible. All the classrooms are accessible, but not all dining rooms are accessible." For three years, Tayjus has advocated for another card reader to operate the elevator in his dorm. To date, his attempts have been unsuccessful. "I wait at the elevator—sometimes for 15 minutes—for someone to come by and press the button," he says, admitting his frustration at how unresponsive the university has been to his needs.

Making friends at Harvard was also difficult. The stigma surrounding disability was real. "People were nice, but interactions were forced, not meaningful." Accessibility compounded the problem. "If I made a friend in another dorm, I usually couldn't visit them because their dorm was not wheelchair-friendly. If people said, 'Let's go out to lunch!' and I couldn't go, I'd come back to my room. I was trapped in my dorm. Alone."

Tayjus' expression changes from a furrowed brow to a wide charming grin when he talks about the friends he eventually found. "I made a friend group and they were mostly international. One of my really good friends is from Uganda, another is from Pakistan, and one is from the country of Georgia. One guy, who is my best friend, picks up on my needs. He zips my jacket and wipes food if I spill. Other people have seen that and learned from him how best to help."

At the foundation of what makes Tayjus' life function is his dedication to keeping himself healthy. As is typical in men with DMD, his lungs are a fragile ecosystem, susceptible to complications from the common cold. Respiratory infection can be fatal. During his second semester of freshman year, Tayjus contracted pneumonia. With fluid-filled lungs and a raging fever, he was bedridden for two weeks and required three rounds of antibiotics. "The doctor said my lung function was a lot lower than it used to be. He said I had to use a cough-assist machine twice a day, even when I was not sick." To demonstrate, Tayjus asks for the mask to be placed on his nose and mouth. The machine sounds like a vacuum, forcing him to inhale first, and then suction out a powerful cough, to dislodge any phlegm or mucus built up in his lungs. "It's hard to cough on your own with Duchenne. I do this once in the morning and once at night. If I get sick—the minute I get a scratchy throat—I use it aggressively." He admits he gets scared when he sees older men with Duchenne who are trached or wear a BiPap (or another breathing machine), full-time. "I do whatever I can to preserve my lung function."

Part of Tayjus' advocacy involves speaking with newly diagnosed DMD families. "When a child is first diagnosed with Duchenne, the parents think about their kid not walking or playing sports. They are worried about how long their child will live." He reminds them that this is the best time for a kid to be born with Duchenne because there are so many options. He tells people about the dozens of companies that are working to improve people's lives through the development of innovative therapies, and the creation of devices to help support the health of people living with DMD. "It gives them hope," he says.

The ideals of biotech and pharmaceutical companies resonate with Tayjus' professional goals: to extend and improve the lives of boys and men living with DMD.

"I made a friend group and they were mostly international. One of my really good friends is from Uganda, another is from Pakistan, and one is from the country of Georgia. One guy, who is my best friend, picks up on my needs. He zips my jacket and wipes food if I spill. Other people have seen that and learned from him how best to help."

He plans to work in public policy, lobbying for access to care for all individuals with Duchenne. Already, he has worked closely with Parent Project Muscular Dystrophy, having served on several advisory boards and advocating on Capitol Hill. He also has brought a patient perspective to a number of pharmaceutical developers, working as an intern for two companies who are researching drugs for DMD. When asked if he'd ever consider running for office, his eyes hinted at a sparkle. "I don't know... Maybe!" Today, Tayjus sees the many challenges ahead of him as doors he has yet to pry open. With his education, work as an advocate and his personal experience living with DMD, he is uniquely poised to unlock each one in due time.

"I would say what guides me is always maintaining a positive attitude and remembering that my disability can be my greatest ability. Sometimes, a disability can lead you to focus on what you can't do, but I believe in maximizing what I can do. It's an important way of dealing with my situation in an effective way."

Siddharth

Born May 1995 | Cupertino, California
Living with DMD

Siddharth's day starts before the sun rises, when his father wakes him and starts the laborious task of getting him ready for school. "I need to get up early to get him to the bathroom first," describes Satya, Siddharth's father. "The whole process takes almost three hours. I don't get hung up on that. I just get ready and go."

One crucial tool for moving Siddharth is the ceiling-mounted track lift which traverses from Siddharth's bedside to his closet, his bathroom, and the outside hallway. Satya carefully connects the straps to support Siddharth's weight, and with the push of a button is able to safely transfer him to his power wheelchair. Satya manages much of the morning's tasks with the help of this machine and his own strength. Bracing his lower back, Satya uses a pivoting motion to move Siddharth from his stander (a therapeutic standing apparatus used for stretching) back into his chair, utilizing momentum to successfully complete the transfer.

Siddharth and his family endure this routine Tuesday through Thursday, in order for him to get to class at San Jose State, where he is finishing up his

bachelor's degree in communication studies this spring semester. He has his sights on a master's degree in the coming years. "Before starting in communication studies, I only just discovered my passion for writing," says Siddharth. "Now, my aspirations are to work in the media industry, for NASCAR, as a reporter writing relevant articles about each race throughout the season." For an adult living with Duchenne muscular dystrophy (DMD), these sorts of trajectories represent a new chapter in the story of the neurodegenerative disease, which once predominantly robbed young men of the ability to ever consider having a career. Siddharth's story is full of inspiration for young men with the disease, who undoubtedly hold similar aspirations and capabilities.

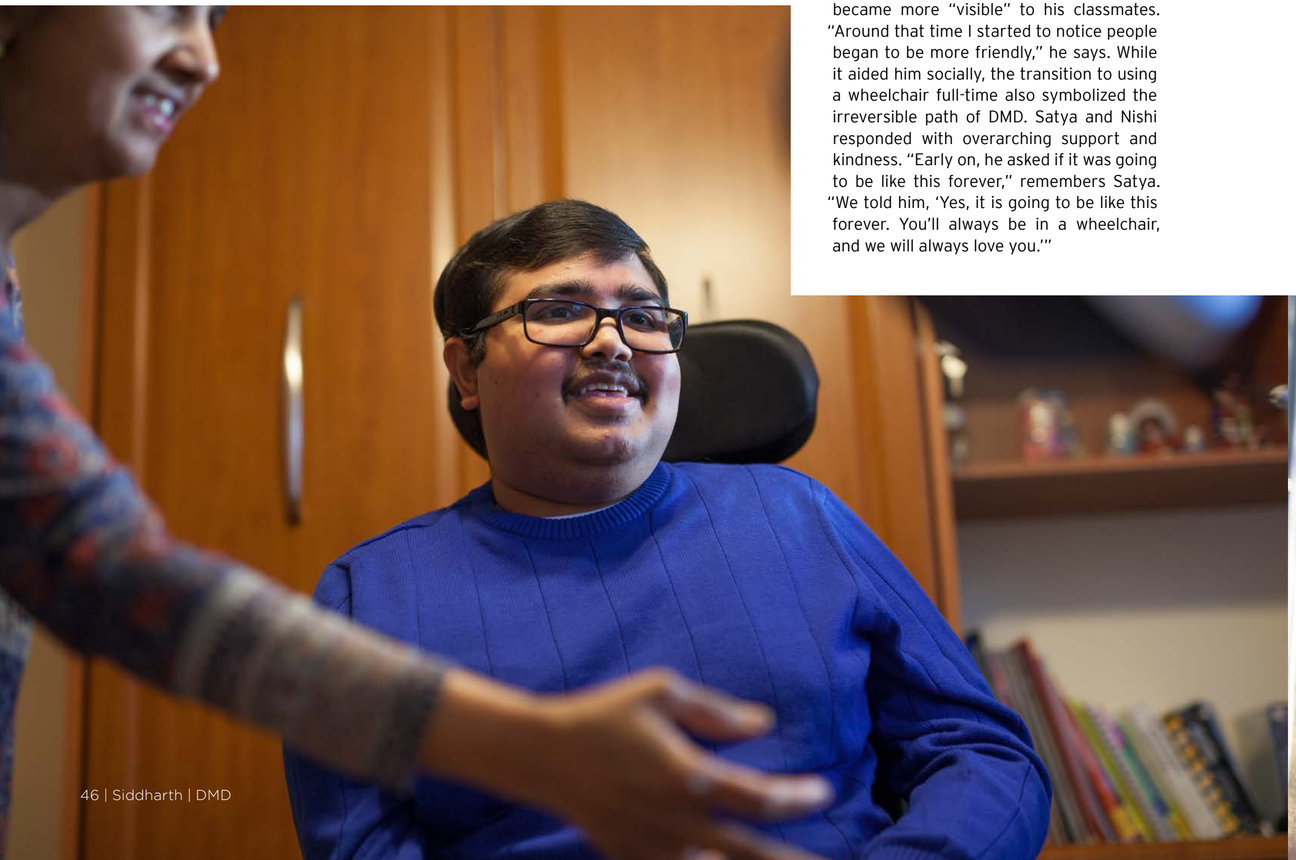
Siddharth was born in Hyderabad, India, but moved to the California coast at the age of one. Satya and his wife, Nishi came to the US for economic opportunity and for the chance to live near family. "I worked in IT," recalls Satya. "I wanted to come here, and my job agreed." Nishi had extended family in California. "I have nine siblings," she says. "And seven of them lived in the US already. We were actually the last ones to come here."

Around the age of five, Siddharth started to show early signs of DMD. "In 1999, he was about to go into kindergarten," remembers Satya. "He was at summer school and the psychologist noticed that he wasn't walking straight—that he was not keeping up with his peers—and told us to take him to a neurologist." The neurologist ordered a blood test for Duchenne. Within weeks, the family's future was forever changed as they learned that Siddharth had been born with a severe, life-limiting disorder, with few treatments and no cure.

"In the beginning, I was not ready to accept it," says Nishi. Neither of his parents had heard of DMD and they couldn't bring themselves to talk about it openly, even with other family members. "It took several years to speak to other people about it," says Satya. "For me, at least, it took a lot of time." Nishi and Satya grappled with the implications of the disease within their family and community, but ultimately responded proactively, seeking resources and guidance from the Muscular Dystrophy Association (MDA). "We got this book [A House for All Children] that helped us redesign our house," explains Nishi, pointing out the wider doorways and hallways. "That way, we were ready for the future."

For Siddharth, realizations about his condition came with his diminishing physical abilities. "I gained an understanding of it a few years later, when I started to see that there were a number of things I could not do at the pace of other kids." He struggled to make friends and found himself socially isolated. "That was another frustration for him," says Satya. "He would correlate that with his diagnosis. He would say, 'Because of my diagnosis, I don't have any friends.'"

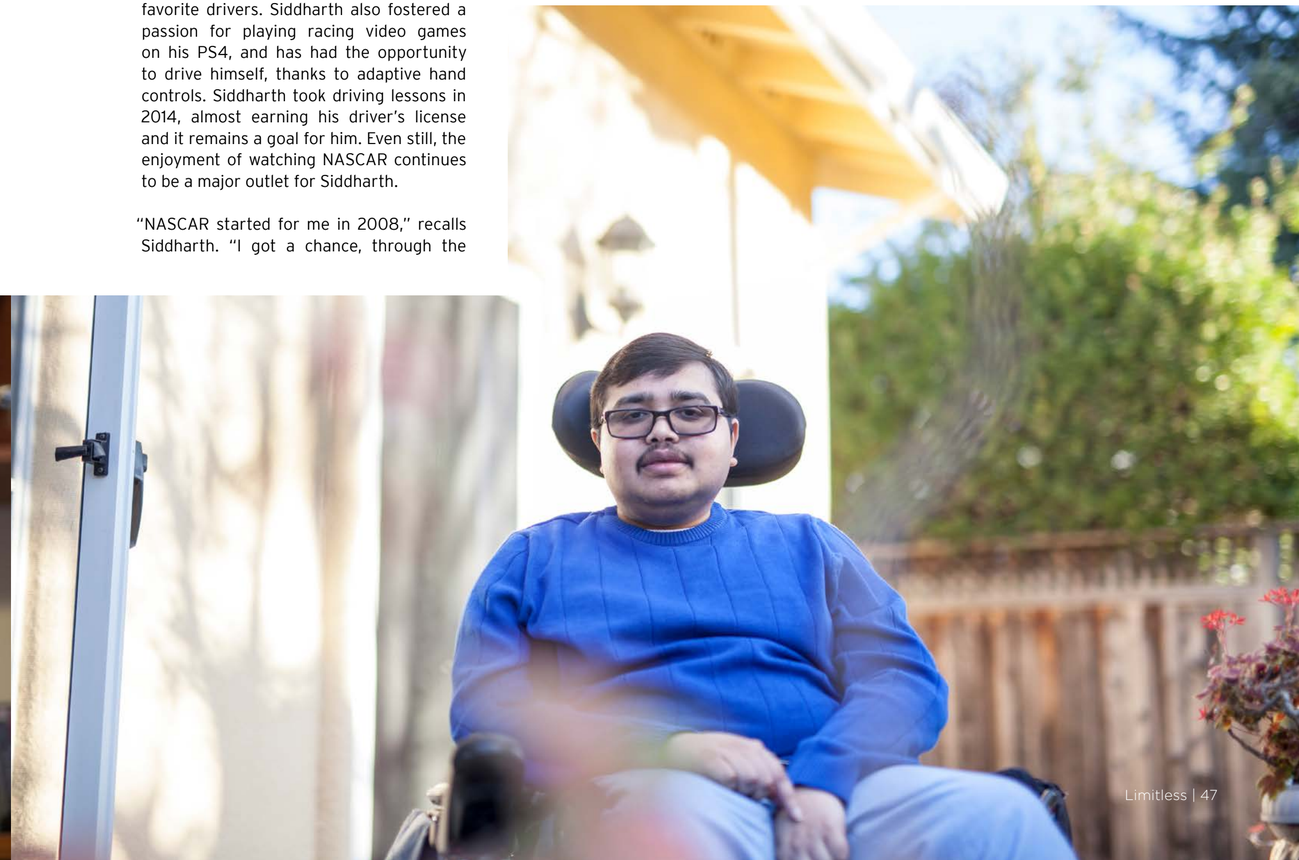
The struggle for acceptance, both of his condition and amongst his peers, was central to Siddharth's adolescence. "When I started to use the wheelchair full-time—that is when I started to adopt a more positive outlook. It built my confidence, and I was able to make a few more friends." Likewise, teasing and bullying lessened as Siddharth's condition became more "visible" to his classmates. "Around that time I started to notice people began to be more friendly," he says. While it aided him socially, the transition to using a wheelchair full-time also symbolized the irreversible path of DMD. Satya and Nishi responded with overarching support and kindness. "Early on, he asked if it was going to be like this forever," remembers Satya. "We told him, 'Yes, it is going to be like this forever. You'll always be in a wheelchair, and we will always love you.'"



Siddharth accepted his reality, and at the same time, found ways to embrace it. From his earliest years, Siddharth held an infinite fascination with automobiles: from miniature toy cars and trucks, to emerging technologies, and professional-level competition. He began dreaming in circles. Images of race cars careening around a cambered tarmac—rubber melting onto pavement, roaring engines, soaring speeds—fueled an unquenchable curiosity. Now, each weekend, Siddharth sits with his family and friends and watches live NASCAR races on the family's big-screen TV, cheering on their favorite drivers. Siddharth also fostered a passion for playing racing video games on his PS4, and has had the opportunity to drive himself, thanks to adaptive hand controls. Siddharth took driving lessons in 2014, almost earning his driver's license and it remains a goal for him. Even still, the enjoyment of watching NASCAR continues to be a major outlet for Siddharth.

"NASCAR started for me in 2008," recalls Siddharth. "I got a chance, through the Make-a-Wish Foundation, to meet driver Jeff Gordon." This piqued his interest, and along with Satya, the father-son duo began watching and attending races, once traveling to the famous Indianapolis Motor Speedway. "Jeff Gordon was my favorite driver for a while, but he retired in 2015," says Siddharth. "Now, I root for Chase Elliot." Satya is quick to rebuke. "Jimmie Johnson is the best driver. I root for him." He pumps his fist in the air in an unexpected jeer of excitement, from the typically mild-mannered gentleman. Their rivalry is alive and well.

Likewise, power soccer, a sport that highlights the abilities and skills of power wheelchair users, came into Siddharth's life in his late teenage years, providing him with an important competitive outlet, as well as a way to bond with others. "It has been a source of empowerment for me," he exclaims. "I view power soccer as a way to showcase my true abilities and uplift



"Developing my oral skills and continuing my education will maximize my abilities, so I can realize my dreams of working in the media industry."

against pessimistic attitudes." He plays on the Steamrollers, a San Jose area team, that travels across California for games, as well as to the national tournament, in Indiana, where the team achieved second place in their division last year.

New passions unearthed new strengths for Siddharth, and he soon fostered a desire to espouse the lessons of living with DMD as a way of radiating positivity to others. "I refuse to let my disability stop me from pursuing my passions and living a meaningful, accomplished life," he states profoundly. "I would say what guides me is always maintaining a positive attitude and remembering that my disability can be my greatest ability. Sometimes, a disability can lead you to focus on what you can't do, but I believe in maximizing what I can do. It's an important way of dealing with my situation in an effective way."

Siddharth speaks slowly, and responds at his own pace—pausing before each elucidating statement—the impact of his deliberate words given time to linger in a listener's ears. While Siddarth is clearly a bright young man, he struggles with speech and language, and he has worked hard to fully develop these skills. As he strives to get into the media industry as a writer and commentator, these are hurdles he is poised to overcome. "The oral aspect is as important as the written aspect. To improve my speaking skills, I am currently attending a local Toastmaster's group." The meetup has given him a chance to practice public speaking. "Developing my oral skills and continuing my education will maximize my abilities, so I can realize my dreams of working in the media industry," he relates.

Writing, too, is crucial to Siddharth as a tool for learning and self-expression. He wheels into the at-home office to start this weekend's assignments. Nishi helps carry some of his books and sets them down on the broad desk. Siddharth is still able to use his hands to type (something relatively uncommon for men his age with DMD), and he makes avid use of this ability. A couple years ago, he started writing for the MDA, starting a blog called "Spreading the Wings of Life," where he muses on life's lessons. Writing and communicating have become a powerful act for Siddharth, and one he can tackle much on his own; a mark of independence for a young man who finds himself increasingly dependent on others.

Knowing the harsh prognosis, the family spends each day focused on caregiving and necessary tasks, avoiding more pernicious questions. "I don't worry too much. I do my routine, and then, all is well," says Satya, intoning his personal mantra. "That's why I don't focus too much on the future and at the same time, I don't think negatively about the future." The coming years will bring new challenges to this family even with the potential for groundbreaking treatments. "I have hope that something will come up," says Siddharth of new DMD research and drug developments. "I don't regularly look at research updates because if it's meant to be, it will work out. I focus on what I can do right now."

Olivia

Born January 2012 | Kansas City, KS
Living with CMD

"Come on! Let's play," exclaims Olivia, her enthused, high-pitched voice trailing behind as she races down the road in her hot-pink power wheelchair. At age six, she's got the energy of a hummingbird, and her rare condition of congenital muscular dystrophy (CMD) does not stop her from going where she wants, at top speed. Olivia lives with CMD LAMA-2, a type of muscular dystrophy related to a specific mutation in the laminin protein. The condition takes two forms: either a severe, early-onset form or a milder form that appears later in life. Olivia's condition was early-onset, though when she was born, her health appeared to be fine.

Olivia was placed in daycare as an infant, as her parents, Matt and Sara, both worked full-time in finance and marketing. During this time, the differences between Olivia and the other children began to appear. "She didn't have the same forcefulness of movements and projection of cries and screams as the other children," recalls Sara. "I wasn't sure what was going on, but my mother's intuition said, 'Something's not right.'"

When Olivia was three months old, Matt and Sara embarked upon the difficult odyssey of finding a diagnosis. "Most people do a muscle

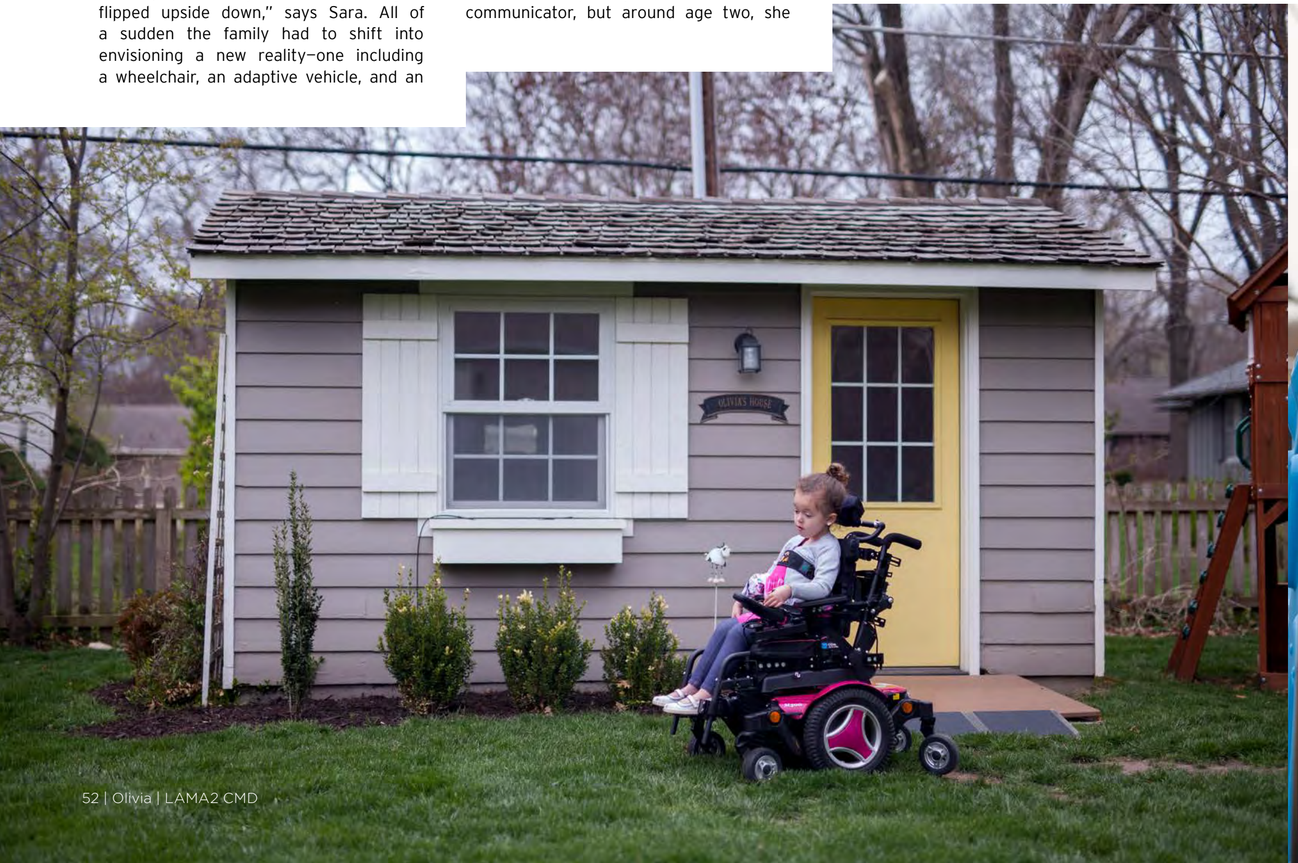
biopsy, but we felt it was very dangerous to give her anesthesia," says Sara. They preferred to get genetic confirmation through blood tests. The genetic testing revealed that Olivia had a high creatine kinase (CK) level, indicating muscle damage. After waiting weeks for the blood results and additional testing, Olivia was officially diagnosed with CMD LAMA-2 at six months old.

Receiving the diagnosis for their first-born baby was surreal. "You think everything's normal, and then your whole world gets flipped upside down," says Sara. All of a sudden the family had to shift into envisioning a new reality—one including a wheelchair, an adaptive vehicle, and an accessible house. Up until that point they had planned on living, working, and raising Olivia in a fourth-floor, walk-up apartment in downtown Chicago. Reckoning with the change in plans, Matt and Sara decided to sell their apartment and move back to Kansas City, where they could be near supportive family and find a house that would accommodate Olivia's needs.

Although Olivia's diagnosis came early, at times it was deceiving, as she showed to be advanced in many ways. Not only was she a very early talker and excellent communicator, but around age two, she taught herself how to read. "Her reading was wild," remembers Sara. "She literally learned to read, self-taught, out of the blue. She memorized words immediately."

As Olivia grew older, Matt and Sara realized that she had no arena to explore her burgeoning imagination and her unquenchable desire to play. They wanted to build her an accessible playhouse, but were met with legal restrictions. "Living in a historic neighborhood, there are many building regulations and constraints," explains Matt. "There was no playspace for her. Not a spot for her." Olivia couldn't go to her friends' houses because they weren't wheelchair accessible, and due to housing regulations, the family wasn't allowed to build another structure big enough for her to have a playhouse.Taking matters into their own hands, they organized a meeting with the building commission, where Olivia, at just two-and-a-half years old, showed up in true form. "In the middle of the meeting Olivia started beeping the horn in her chair. 'I'm bored, I'm bored, I'm bored. I am so bored,' she exclaimed in her sweetly adamant voice. 'I have nowhere to play! Can I just have my playhouse?'" After witnessing young Olivia, the building commissioner approved, and the family was permitted to build a playhouse large enough for her to play in.

"Olivia is unique because she is so verbal," reflects Sara. "Olivia speaks up. We do this for Olivia and for all the other kids too." Furthering her knack for civic engagement, Olivia helped redesign a playground in her neighborhood. The plan for the playground was to have mulch and woodchips on the ground beneath the structures, in which case, Olivia wouldn't have been able to access it. So she spoke out to be included, rallying for a soft rubber surface instead. "Our neighbors were really great about standing up," says Sara. "They'd say, 'We need to be looking out for kids like this.



We need them in our neighborhood.'" The communal camaraderie moved Sara to tears. "People care, but they don't know what they don't know. Once we started talking, we've been surprised by the support we've been given." With this support, Olivia's family was able to accomplish major milestones with accessibility to public spaces, and not just for Olivia, but for other children as well.

Managing Olivia's medical needs have also proved challenging. "We are lacking an integrated team," explains Matt. "The doctors are good but they don't communicate with one another. Instead of having a neuromuscular clinic, for instance, we have to be the communicators between specialists." This lack of integration takes extra time and is a cause of frustration. At some point in the future Olivia will need a spinal fusion. But it's really when her spine will start impacting her lungs that concerns Matt and Sara the most. They will have to go back and forth from pulmonologist to surgeon—requiring precious time and energy—to figure out what is best.

As she speeds around the house in her pink wheelchair, her little sister, Emily, hops on for a ride. Emily sneaks a sip out of Olivia's water cup that's held behind the seat. Matt and Sara catch it and quickly move to the kitchen sink to wash the cup. "It takes her three times longer to get over any illness," remarks Sara. "So we have to be really careful she doesn't share with other children to keep her healthy." When Olivia was two, she contracted pneumonia and was taken to the hospital where they found that half of her lung had collapsed. She was put on respiratory-support for a week. Two glucose crashes also led to more hospital visits. Through observation by a feeding specialist, they discovered that every time Olivia was taking a sip of water, she was silently aspirating—breathing in the water. "The feeding therapist was a saving grace because after that, she started doing so much better," recalls Sara.

Time is the main ingredient in caring for Olivia. "It used to take us an hour-and-a-half per meal to feed her. Doing that three times a day takes almost a whole day." With the support of Olivia's grandmothers, nannies, nurses, and specialists, the time obligations are shared. Sara's mom has been the biggest help with babysitting and feeding, but soliciting her help was easier when Olivia was smaller. "Our moms are older now and Olivia's getting heavier, so it's difficult to find people who can pick her up and transport her in a way that they are safe and she is safe," explains Sara.

Despite their support network, Matt and Sara still find themselves missing work quite often for illness, doctor's appointments, IEP meetings, and swimming for physical therapy. "It's a different level of tired that we operate at," says Sara. And step by step, they are always learning. Their greatest resource is connecting with other families online. "Even our family members don't really understand on a day-to-day, hour-by-hour basis what's going on. If we didn't have access to other families with CMD-LAMA2, I don't know what we would do," expresses Sara. "Connecting with other families is critical. They can give you a glimpse of what's next. We rely on them more than anybody else. There's so much more information I can get from them, more than doctors. That helps with not being so overwhelmed."

Matt and Sara try to focus on the areas they can help. Sara assists with fundraising and social media efforts for the Cure CMD organization. "You do feel helpless," says Sara. "There's so much you can't do. We can't fix it or change the diagnosis, but we can change certain things to make her life and others' lives better. That helps." Olivia's resilience in itself is an aid. She has been a spokesperson for organizations like the Variety Children's charity, a national organization with a Kansas City chapter. Olivia has appeared on commercials for the local grocery store, and even at cinemas across the midwest, on an advertisement before each film supporting Variety's efforts to build inclusive playgrounds. Her lively spirit brightens up any room. "Being around Olivia makes my day better," says Sara, with a smile.

A social butterfly since she was very little, Olivia is always talking to people. "She'll go zip up to all the neighbors, or anyone, and talk," says Matt. When people ask Olivia about her wheelchair, she replies, "My muscles don't work, so I have a wheelchair that helps!" For Olivia, it's as plain and simple as that. "Kids should be able to be included and be able to play with their friends," says Matt. "It's all stuff we never thought about before, and a lot of people in the community don't think about either, until they meet Olivia, and see her firsthand."

Matt and Sara keep up with the hurdles of learning how to best care for their daughter. "We need her to be here, and we need her to be healthy. And healthy doesn't mean you have to walk." They are focusing now on her spine and breathing support, to keep her lungs strong. "She's currently very healthy, in good weight. We want her to maintain independent eating and breathing," says Sara.

Olivia picks out another color from her set of fifty markers. "Will you pass me eggplant purple please?" she asks. She knows the full name of every color, and rotates between ten as she fills in the gemstones on a paper tiara. When asked to pose for a picture, she obliges, but only for a moment. "But I'm coloring! Can I go back to my very important coloring now?" On the one hand, Olivia is focused on typical six-year-old stuff—making art, playing with friends, and playing games. But she also does the extraordinary—she speaks out about her condition and advocates for herself. "We talk about resilience. Olivia is the epitome of it," says Matt. "She's the most resilient kid, and human, I've ever been around."

"Being around Olivia makes my day better."

–Sara

Chaz

Born September 1993 | Littleton, CO
Living with CMD

"I want to educate the larger community. I want people to see visually impaired people as assets, not as helpless."

Wearing shiny blue running pants and a black Team USA jacket, Chaz Davis walks across the parking lot, the ground covered in patches of hardened snow. Ice cracks beneath his feet as his white cane swishes softly in front of him. He arrives at the track and bends down, letting his cane drop to the ground. "Running has always been a huge part of my life."

Chaz is a competitive runner, holding state titles, medals, and dozens of team championships. As a freshman at the University of Hartford, he ran cross country and finished the year as the team's top freshman runner. He has run in the Paralympics and most recently the California International Marathon—all without being able to see.

Chaz was born with perfect eyesight. One morning his freshman year of college, his vision suddenly changed. "I woke up one day and my right eye couldn't see at all," Chaz says. "My vision was distorted. Light was coming through with some peripheral vision, but my central vision was gone." He assumed the vision change was associated with recurrent migraines,

but within a week, the vision in his right eye had further deteriorated. The first round of medical tests revealed no answers. It wasn't until a few months later, when the vision in his left eye deteriorated, that Chaz met with experts at Tufts University.

A neuro-ophthalmologist told Chaz he had Leber hereditary optic neuropathy. LHON is a rare mitochondrial disorder that causes degeneration of retinal cells which can lead to a sudden-onset loss of central vision. It affects one out of 50,000 people. Chaz's particular mutation, 3460 (the point of mutation in his mitochondria), affects his peripheral vision as well. Additionally, he has what is known as LHON Plus, a more severe form of LHON which involves extraocular conditions—in his case, neurological conditions like muscle twitching and fatigue. Although hereditary, no other members of Chaz's family had any LHON symptoms.

"I thought the doctor might say I'd get my sight back," says Chaz. "But he took my parents into his office. When they came out, they were very emotional. They told me that I would be legally blind for the rest of my life. That summer, I began drinking. I went into my room and turned the lights off. I didn't want to do anything."

Chaz describes what's left of his vision in two parts. First, he describes his central vision as a screen-like field of pixelated dots. Occasionally, he can detect blurry images in front of him that are blue or brown. Second, his remaining peripheral vision consists of narrow slits in which he can make out rough images, such as three fingers he holds out an arm's length from his ear.

A few months into his self-described period of "self-pity," Chaz woke up and thought I'm not doing this anymore. "Somehow, I got my spark back," he says. Newly blind, he returned to school. "Within a week of arriving, the Massachusetts Commission for the Blind (MCB) set me up with computers and assistive devices. But the commission representative warned me that I shouldn't go back to college—that I wasn't ready—and that if I tried, I would likely fail." Chaz disagreed. "With all due respect, sir," he said, "I'm going back to school." He found a tutor to teach him about software that converted text to audio files and excelled, earning a GPA of 3.8 that semester, his best up to that point. Years later, when Chaz graduated, his college rehabilitation counselor photocopied his diploma and mailed it to the MCB. The message was clear: Chaz did not accept failure as an option.

"The commission representative warned me that I shouldn't go back to college—that I wasn't ready—and that if I tried, I would likely fail. 'With all due respect, sir,' I said, 'I'm going back to school.'"

Although Chaz was succeeding academically, he missed competitive running. He longed to be a part of a team again, and to feel once more the physicality that was his natural born gift. He was unsure if there was a way to run as a blind person.

The Grafton Gazebo Road Race was a local five-mile race that Chaz had won in the past. The community heard of Chaz's story and decided to donate proceeds raised by the race to fund LHON research. "If they are going to do that," Chaz thought, "I at least have to try and run the race. At that point, running five miles was way out of the question," he laughs. "Five miles! Are you kidding me?"

"My friends from school came to guide me in the race. I had gained 55 pounds; so here I was, this bloated guy trying to run five miles around town. I placed fifth, but more importantly I was able to cross the finish line with my friends. That changed my perspective on everything. I knew I had to get serious about running again, but I had no idea how."

Chaz trained on a treadmill, starting slow and then cranking it up to six-minute miles. "That fall, I tried to run the preseason trials with my team, but I fell down a bunch of times, tripping on rocks and sticks. It was really disheartening. I felt like I was in good shape, but I had this barrier." Chaz's coach suggested he give indoor track a shot because of the smooth running surface. It was an idea that worked. "With my peripheral vision, I could discern the lines of the track. I didn't fall at all. I could innately sense other runners around me." Running track gave Chaz a boost in confidence. He increased the intensity of his training and soon became the fastest runner on the team.

The following summer, Chaz had a thought: "If I'm blind and running, there have to be other blind runners out there. I want to compete against people that are similar to me." He heard about the Paralympic games and got in touch with the coaches for the United States team. The national trials were just one month away. "I said, 'Well, I'm going!' Me and my parents hopped in our car and drove straight to Minneapolis. I ran the 5K and won. I qualified for the team."

Back at school, a new coach was hired. Coach Roger Busch was excited to train Chaz for the Paralympics, but he also wanted Chaz to return to cross country. "I just looked at him and said *'What?'* Cross country is trails and uneven terrain. How is that gonna happen?" Coach Busch insisted and a teammate guided Chaz, alerting him to obstacles along the way. For the first time since he became blind, he felt the rush of running past trees and the textures of earth beneath his feet. Chaz says, Coach Busch "was the first person to not mention my blindness as a defining characteristic of who I was."

The 2016 Paralympics in Rio de Janeiro were on the horizon. Chaz would be competing on an international stage with other visually-impaired runners from around the

"Don't call me inspirational if I'm walking across the street. Everybody walks across the street. If someone who is blind finds me inspirational for running, or getting a degree, that's okay. If I can inspire someone just by being myself, then that's good."

world—people who struggled like him and trained like him. But the morning of the first race, his neurological symptoms flared up. "I was extremely fatigued and I had muscle twitches. I was really worried." When Chaz arrived at the stadium, however, he heard something that gave him new-found energy. "It was the sound of 50,000 people. There was this huge roar. You could not hear your feet on the ground. It got me going!"

Chaz speaks with pride about representing his country on the international stage. He also laughs about how unlikely it all seems: Three years after forcing himself out of the darkness of his bedroom, he set an American record, placing 10th in the 1,500m at 3:58, which is roughly a 4:15 mile. He also placed eighth in the 5,000m at 15:15.

Coming off the high of the Paralympics, Chaz says it was a challenge to adjust to everyday life. "I kind of realized then… I'm actually blind. The world around me is a sighted world. It's not set up for blind people to succeed. I needed more [life] training." He moved to Denver and enrolled in the Colorado Center for the Blind. There he found a community of blind friends and role models. The center taught independent life skills like Braille and cooking. With hearty laughter, Chaz recalls the day he mastered navigating public transit. "One day the instructors put us in a white van with blindfolds on. They drove all over the city and brought us to some undisclosed location and said, 'Ok, get out!' I had to find my way back to the center. I stood there terrified for two minutes. I heard traffic to my left. On the sidewalk, I tapped my cane on a pole. I could feel it was a bus stop. I took several buses, listening for the stops until I recognized the downtown area. Because I could see before, I could visualize the route like an internal map. That kind of independence was new to me."

Chaz recently enrolled in a masters in social work program at the University of Denver, where he is pursuing clinical social work within the blind community. He also recently made his marathon debut at the California International Marathon running it at 2:31:48, setting an American record in the visual impairment category. Chaz insists his accomplishments are more than just personal successes. "I want to educate the larger community. I want people to see visually impaired people as assets, not as helpless. I am a representative of the blind community whenever I'm in public."

Asked if people call him inspirational, he laughs. "Every single day! But don't call me inspirational if I'm walking across the street. Everybody walks across the street. If someone who is blind finds me inspirational for running, or getting a degree, that's okay. If I can inspire someone just by being myself, then that's good."

"My biggest goal was still having the perception that I could do anything. Whether I actually could or not didn't matter. I still wanted to have the perception that I could."

Jessica

Born March 1986 | Tucson, AZ
Living with LHON

With all her belongings packed carefully into boxes in her parent's garage in Phoenix, Arizona, the day was fast approaching for Jessica to close on her house. At age 27, she was newly employed with her first "real job" out of college. She always dreamed of buying her own home and had finally found one she could afford. But just as she was finalizing the paperwork, a problem arose. Her vision had changed; she couldn't see out of her left eye. "I was just praying it wouldn't go to my other eye," recounts Jessica. Symptoms did not let up, however, and within a few months the vision in her right eye started to blur as well. Jessica called her mortgage broker to convey the troubling news. "Listen," she said hesitantly. "I can't go through with this. I'm losing my vision." Jessica had to back out of buying the house, and began the reverse process of unpacking all of her boxes and moving back in with her parents.

“‘Hi my name is Jessica. I have a brand new job and I think I’m losing my vision. Can you help me?’”

Within six months, Jessica went from being a fully sighted, fully functional, independent young woman, to becoming severely visually impaired: unable to recognize faces, stairs, or written words. “How will you cook without chopping your fingers off?” her mother had exclaimed. Jessica spent the next year-and-a-half living with her parents where she re-learned how to cook, clean, communicate, and traverse the world with a blind cane.

During childhood, her younger brother had lost partial vision at the age of eight. “Back then all it meant was that his homework was blown up really big and he sat close to the TV. We would go to doctor after doctor, but no one knew about LHON at that time.” LHON, or Leber's hereditary optic neuropathy, is a mitochondrial inherited degeneration of retinal ganglion cells (RGCs) and their axons that leads to a loss of central vision. Jessica’s aunt also went completely blind at age 62. Jessica noticed this pattern in her family, but when she first lost vision in her left eye she refused to believe there was a hereditary connection.

“Hi my name is Jessica. I have a brand new job and I think I’m losing my vision,” Jessica spoke into the phone. “Can you help me?” On the receiving line was Lissa, a woman Jessica found through her initial searches online. Lissa administered a Facebook page for the LHON blind and visually impaired community, a role she took on after her own son was diagnosed with LHON. After a quick message response to Jessica, Lissa became an invaluable friend and mentor. “Lissa was there for me immediately. She told me everything I needed to know and

what I needed to do and learn at that time." Through Lissa's Facebook page as well as Arizona Services for the Blind, Jessica found excellent resources for training and support. Jessica's searches online also uncovered many glaring stigmas associated with blind and visually impaired people. "There is so much depressing stuff about blindness, like the stereotype that blind people always look disheveled; that they just roll out of bed with sunglasses on." Wearing an immaculately ironed silk dress, high heels, and expertly applied makeup, Jessica clearly defies any existing stereotypes.

This took no small amount of effort. "My first major hurdle was still looking put together; like I hadn't missed a step." Although Jessica cannot recognize faces, she converses loquaciously while looking you straight in the eye. "I tell people that I see *underwater*. My world is really blurry—I have no central vision. I can't see your face, but I know that you are there."

With such stark loss of vision, Jessica could no longer drive, making commuting the next challenge in her desire to be an independent woman with her own career. "I knew it was no longer safe [to drive] when the halos were so big around the lights that I didn't know if they were green or red. That's when I knew I should be off the road." When Jessica sold her car, it marked a significant turning point in her independence. "Everyone with LHON remembers the last time they drove a car."

Despite transitioning to paratransit, Jessica never lost her drive to remain a fervent young professional woman. One day, while catching the Sun Van (Arizona's paratransit), the driver exclaimed to her, "What? I thought you can't see—what are you doing wearing heels like that?" To which Jessica replied, "My legs aren't broken!" The exchange inspired Jessica to start her own blog, appropriately titled *Blind Girls Can Rock Heels*, where she describes her experience living with LHON and deconstructing the barriers associated with visual impairment. "My biggest goal was maintaining the perception that I could do anything. Whether I actually could or not didn't matter. I still wanted to have the perception that I could."

When Jessica first lost vision, it was imperative to her that she keep her new job. But how? It was something she could not do alone. While keeping up the impression of a fully sighted, fully functioning Sales and Marketing Coordinator, every evening after hours, her mother, father and best friend would sneak into her tiny office to help play catch-up. From about 8-11:30 p.m. they worked—her best friend was the typist, her mother would file and her father would organize. "Then we all went home, and I came into the office the next morning saying, 'Oh that was so easy!'" For months, unbeknownst to the company, Jessica's family and best friend helped her with the basic tasks she hadn't yet mastered since losing her vision that were needed in order for Jessica to keep up her employment. She kept the coffee brewing for her secret helpers as she slowly learned how to efficiently multitask without dependency on her eyes to guide her. "We spent hours catching up, and no one knew." Jessica proved capable of learning to do her job without vision and although she required some extra assistance, she began to grow her nascent career in marketing and public relations. Years later she would land a job at the University of Arizona in the Clinical Trials Department, moving from Phoenix to Tucson—her "happiest place on Earth."

As her vision changed, so did her hobbies and friends. Jessica began to meet other people within the LHON community and was especially inspired by a young man named Chaz, who despite his vision impairment, was an accomplished long distance runner. He challenged her to try running and she loved it. "I was never a sighted runner," she distinguishes, as it wasn't until after she went blind that she started. "I had no idea what blind running meant. It was the scariest

thing at first." To start, she connected with a few running guides who one by one took her out on simple trails. Her mother would meet each of them and wait in the car for their return, until they proved trustworthy. "When everyone asked what I was training for, I told them I was just a happy runner. I didn't have to use my cane, or use my vision, I just got out there and ran free. Whenever I'm running I have a huge smile on my face." What LHON had robbed of her sight, running returned of her freedom.

With a new outlook, Jessica started organizing Dinner in the Dark fundraisers, in which sighted participants share a three-course meal completely blindfolded. The first dinner she held in Phoenix was advertised all through word of mouth and welcomed 98 people in attendance. Her second year brought 143 participants, including congressmen and firefighters, and during her fourth year, she raised the most money yet. In total, Jessica has raised over $25,000 for the blind and visually impaired community.

Aside from raising funds, the dinners are an opportunity for Jessica to teach the broader public important lessons about the ways blindness and vision are perceived in society. "I wait for that hum of silence when everyone can't find their food," she says. "All of a sudden you hear the hum gets quieter, and that's the moment that I wait for. That's success, you know, the realness sets in." She provides space as well as a platform for the visually-impaired to teach their simple truths—to unmask their condition and foster mutual understanding between the two groups. "I wanted to do something to make a difference, to be able to help do my part. Raise awareness."

After sitting with Jessica outside of her office on the University of Arizona campus, we take a drive into the hills of Gates Pass. Opening up to a limitless vista of the Tucson mountains, we park and then guide Jessica along the dirt path and venture into desert hills. "It's so beautiful out here!" Jessica rejoices. When asked if she can see the plants and the colors, she responds, "I see the blue sky and I see the horizon and yeah, that's all I need." We take a picture with her smart phone; she then holds the phone an inch from her eyes, and while zooming in, alights at the beauty seen there.

As the sun sets over the mountains, Jessica tells us that she will wake up at 4:30 a.m. "to run a quick eight miles before work." She currently runs with guides five days a week and is training to run the Boston Marathon with the United States Association of Blind Athletes. It is remarkable to witness Jessica's strength, perseverance and positive attitude. After becoming severely visually impaired at age 27, Jessica excels in her professional life, lives on her own, runs marathons, and has become an inspiring keynote speaker. Jessica is especially enthusiastic about being a positive role model for younger people and is looking forward to making her keynote speech at ENVISION, a camp for blind and visually impaired children in Wichita this summer. "There's this perception that just because you can't see, you can't, can't, and can't." If one thing is for certain, Jessica can, does, and will. After receiving her diagnosis of LHON, she has taken off running.

About

Living in the Light™ is a patient advocacy initiative utilizing the potency of fine art photography, compelling personal narratives, and engaging filmmaking to educate the biotech and medical community about the realities of rare diseases and the unprecedented effect they have on families and daily life.

This distinct concentration grew out of an understanding that the rare disease community is comprised of many people and many voices. By shifting the focus from patient to person, Living in the Light presents the dignified and sincere perspectives of individuals and their families as they approach major life challenges. Our work is not simply to record, but to convey the deep resilience, unique wisdom, and profound beauty of people as their journeys unfold.

No matter who the patient is, it is the entire family that receives a diagnosis—everyone is affected in some way. We understand this, and our approach is poised to honor and accommodate the families we document. Since our founding in 2012, telling rare disease stories has been our sole focus. It is what we are most passionate about and what we do best. To date, we have interviewed and photographed more than 300 families, living with over 50 different rare conditions around the world.

We are driven by our mission of empowering families and individuals affected by rare diseases to be seen and heard as they relay their stories and advocate for their needs. In doing so, we help our partners form lasting relationships with these families, patient advocacy organizations, and other industry leaders who are dedicated to developing new therapies with the potential to treat a broad range of rare diseases.

To find out more, visit:
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