My Life

By: Tiffany House

The week before my twelfth birthday my life changed forever. It changed when I was diagnosed with Acid Maltase Deficiency Disease (AMD/Pompe Disease), and my mother was told that I would probably not live to be twenty. That one moment changed my future forever. In a split second I went from being a normal, healthy eleven-year old girl to an eleven-year old girl with a deadly disease. Things would never be the same again.

Before I get into all of the ways that being diagnosed with AMD changed my life, I need to explain what AMD is. "Acid Maltase Deficiency is caused by a complete or partial deficiency of the lysosomal enzyme, alpha-glucosidase. This enzyme is necessary to break down glycogen and to convert it into glucose. Without this enzyme, glycogen, a thick sticky substance, accumulates in the lysosomes (sacs within the muscle cells) and leads to severe muscle degradation. It predominately affects the heart, skeletal, and respiratory muscles of the patient." There are two forms of AMD; infantile, and delayed onset. Patients with the infantile form do not produce any of the enzyme and are the most severely affected. Their symptoms appear in the first few months after birth and are characterized "by a rapid build-up of glycogen in muscle tissue causing severe muscle weakness and enlargement of the heart and liver. Respiratory and heart complications lead to death by the age of 12 months . . .The delayed-onset form can present at any age. Delayed onset patients produce a minimal amount of enzyme . . . Glycogen build up is not as rapid as in the infantile form but the disease is progressive and can greatly decrease the life span of the afflicted person. In the delayed onset form deterioration of muscle is mainly confined to the skeletal muscles, the diaphragm, the limb-girdle, and the trunk. Respiratory complications are the main cause of death. Delayed onset patients that present symptoms early in life are usually more severely affected and rarely survive past the second or third decade of life. Patients that experience onset later in life generally progress at a slower pace." I have the delayed-onset form of Acid Maltase Deficiency.

From the time that I was an infant I was susceptible to colds, which could very quickly become a respiratory infection, and from there turn into pneumonia. This was the first clue to my parents that there was something wrong with me. However, it was after watching my younger brother and sister grow up that they began to worry. You see, when I was a toddler, I never tried to climb out of my crib, go up the stairs, or things like that. Because my parents didn't have anyone with whom to compare me, they didn't think anything of it. But when my sister and brother came along, they realized how inactive I really was. While I never tried to climb out of my crib, my parents couldn't keep Andrea in hers, or Randy in his. In addition to that, they were constantly getting into things in ways that I never had. So, they took me to the doctor and told him about their concerns. They were told that I was fine, and that not being really active was probably "normal" for me. They believed the doctors and tried to put their worries out of their minds.

As I was growing up, I never really considered the fact that there might be something wrong with me. I knew that I was not athletic, but neither were lots of other people. I played softball and volleyball. I was on a swim team, and I went to a tennis camp every summer. Even though I was often one of the worst players, I did these activities anyway, and had fun doing them. However, I must admit now, that I had "tricks" that I used so that I could physically keep up with the others. For instance, the swim team that I was on had practices several times a week, and we had to swim lots of laps every practice. My solution was to swim with everyone else, but when half of the swimmers finished and got out of the water, I would get out too. It didn't matter how many laps I had left to go, I would get out. At the time, I would think to myself, "Why doesn't everyone do this? I am so smart!" Looking back, I realize that this was one of the ways that I subconsciously compensated for not being able to keep up with the other kids my age. My mom tells me that I when I was growing up, I would always take charge of whatever group I was in. I would choose the game that we would play, or what we would do. I think that this was another way that I compensated, because it allowed me to choose activities that I knew that I could do. However, despite these subconscious compensations, I never consciously considered the fact that I had something wrong with me. It was not something that ever even remotely entered into my mind. I also had no clue that my parents were worried about me, and I wouldn't until December 1994.

As I got older, my parents continued to worry because in addition to being unathletic, I had also stopped growing correctly. What I mean is that while the other kids my age continued to grow, I had basically stopped. This was of great concern to my parents, and so they started to take me to a lot of doctors. Once again, I had no clue that they were worried about me, and I never really thought about why they were taking me to so many doctors. I mean, when you are a kid (during this time I was between the ages of 8 and 11) you don't question your parents when they tell you that you have a doctor's appointment. At least I didn't. Anyway, all of these doctors told my parents the same thing: "She is normal for her." However, my parents had quit believing the doctors. They knew that something was wrong with me, and they were determined to find out what it was.

Their questions began to get answered in December 1994 when my dad's cousin, Bodie (i.e. Dr. Roy House), came to San Antonio for a medical conference. My parents discussed their concerns with him and asked him what he thought. After having me walk up a flight of stairs and run down a hallway, he told them that he thought that I had a myopathy. After years of seeing countless doctors, he diagnosed me in less than ten minutes! He also recommended that I make an appointment to see somebody at the MDA (Muscular Dystrophy Association) clinic as soon as possible so that we could find out exactly what we were dealing with. However, we could not get an appointment for six months. My parents couldn't wait that long, and so, Bodie got me an appointment at the Mayo Clinic in Rochester, Minnesota for January 1995. I don't really remember what I felt when Bodie gave us the news. All I know is that when I look back at that time, I realize that I had no clue that my life was about to change forever.

The week before my twelfth birthday was to be the week that changed my life forever. I

didn't know this when I got on the plane to go to the Mayo. I didn't know this when I went to have my first of many muscle biopsies. In fact, I wouldn't know this until I was sitting in the doctor's office with my mother hearing the results of the biopsy, and I wouldn't really understand it until years later. It was sitting in this doctor's office that we were told that I had Acid Maltase Deficiency Disease, that I would not live past the age of twenty, and that there was nothing we could do about it. Talk about a kick in the gut. Here I was, feeling like a normal eleven year old, being told that I was going to die in nine years. It was really hard to accept it, and to be honest, I'm not sure that I did. Things changed even more when the results of further testing showed that my lung functions were at 40% of normal. This meant that I was going to have to be put on a Bi-Pap at night. When I take a breath in, the Bi-Pap gives me positive support. What that means is that it gives me the help that I need to take a deep enough breath while I am lying down. It does this through a mask that I have to wear every night, or whenever I lie down for longer than a few minutes. Being put on a Bi-Pap was very hard for me, despite the fact that it is very noninvasive. At first, I did not want anyone but my parents to see me with it on. I felt that if my brother and sister saw me with it on, it would change their perspective of me, and I couldn't handle that. However, I eventually became comfortable enough with the Bi-Pap to let Randy and Andrea see me with it on. Later, I was comfortable enough to let Bertha, our housekeeper/friend, see me with it. But that was it for a very long time. In fact, to this day, the number of people outside of the medical and Pompe community who have seen me with my Bi-Pap on is very small. For me, it is something that is very private, and I guard this privacy very closely. I only allow people that I feel very comfortable with to see me with it on.

Returning to school after the diagnosis was very hard for me. It was the middle of my sixth grade year, and I suddenly had to deal with something that none of my peers would ever have to deal with. Although I had quite a few friends, I did not feel close enough to any of them to tell them about my disease. Instead, I would go to school and pretend that everything was normal. I did this for the rest of my sixth grade year and half of my seventh grade year. However, it was during seventh grade that my health started to rapidly decline. It became harder and harder to get up in the morning for school, and once I was at school, it was hard to walk from class to class. I was on a new diet (that I will discuss later) that made me feel sick all of the time, and then I caught a cold right before the Christmas break. I went back after Christmas, and tried to make it, but things were just too hard. So, in the spring I quit going to class and instead finished up the year by turning in homework that my mother would pick up for me. Because I had never told any of my friends what was wrong with me, I eventually lost touch with them, and from that point on, my friends have been family members, friends of the family, and some wonderful people that I have met as a result of my disease.

From eighth to tenth grade I was home schooled through the NEISD (instead of going to school, the teachers would come to me). My math tutor, Tiffany Huebner, is one of the people that I met as a result of my disease, and she became more than a teacher. She became a friend. Tiffany was a very young teacher who had only been teaching for a few years. We hit it off right away and have been friends ever since. Even when she was no longer my tutor, we stayed in touch and she has been someone that I can talk to about

everyday things. This was really important to me, because it gave me someone, outside my immediate family, to talk to, and that helped to keep me from feeling isolated and alone. Another person that I met as a result of my diagnosis is Jordan Overcash, my personal trainer. In order to stay in as good of physical shape as I could, at the age of thirteen I started working out with Jordan three times a week, and I am still working out with her to this day. Jordan is one of my best friends. Whenever I have a problem, I talk to her about it, and she helps me to deal with it. She has never let me give up, and whenever I get discouraged, she is right there cheering me on. Don't get me wrong, I still complain whenever she comes over because it means that I have to exercise. Whoever said that exercise is fun, and the more you do it the more you enjoy it, is a big fat liar. I have been exercising three times a week for the last seven years, and if I had to do it with anyone but Jordan, I would go crazy. But all joking aside, one of the few positive results of my disease was meeting these two people who have become very good friends, and who have helped me to get through the last seven years, emotionally intact.

However, one thing I am sure of is that I would not be alive today if it weren't for everything that my parents have done. While I was trying to deal with my diagnosis, they would not accept that there was nothing that they could do to save me, and so they started researching AMD. In the course of their research, they discovered that there was research being done to find a treatment for my disease. The research was being done in two separate places, Duke University in the US and Erasmus University in the Netherlands, and it centered on the principle of enzyme replacement therapy, or ERT (basically, in ERT patients are given an infusion of the enzyme that they are missing, or do not have enough of). They also came across a doctor in New York who advocated a high protein-low carbohydrate diet as a way to slow the progression of the disease. They contacted the doctors at Duke and Erasmus and tried to find out how close they were to a treatment. They then formed the AMDA, Acid Maltase Deficiency Association, to promote cooperation between the different scientists in the hopes that there would be a treatment in time to save me. I can't even begin to name all of the things that they have done in the last nine years that have helped to promote a treatment. In short, I believe that it was their determination that enabled a treatment to be available for testing as early as it was and in effect, for saving my life.

While we were waiting for a treatment to be made available, my parents took me to New York to meet with Dr. Slonim and get me on his diet plan. When I was diagnosed, I was only 4'10" and I weighed a mere 58 pounds. Dr. Slonim wanted me to get a feeding tube that would enable me to gain some much-needed weight. However, I didn't want to do this. I was still trying to live as normal a life as possible, and a feeding tube was not in my game plan. My parents understood this and supported my decision. The deal was, however, that I HAD to gain the weight. For the next several years I tried several different diet plans. The first was Dr. Slonim's. He wanted me to eat an enormous amount of protein every day. In order for me to ingest the amount he wanted me to, I had to take a protein supplement (Promod) three times a day. I did this for about a year. However, taking the supplement made me feel sick all of the time. I hated the way it tasted, and I would get very nauseous every time I took it. In fact, I would only start to feel better about fifteen minutes before it

was time for the next dose. My mom finally realized that this was not working for me. I felt like it wasn't worth it if I was always feeling sick. So, she started researching again and, finally, came across the name of a man in San Antonio who also had AMD. She contacted him and discovered that he was on a high fat diet. I tried this, and it worked a lot better for me. I finally started to gain the weight that I needed. However, as I gained weight and grew, I started to develop scoliosis, a curvature of the spine, that is quite common in juvenile onset AMD patients Now, my disease was changing the way that my body looked and as a result my self-image.

As my scoliosis got worse, so did my breathing, and it also became harder and harder to walk. I was at a point where I was going to need major back surgery. However, right around this time (Fall 1998), we learned that the scientists in the Netherlands were almost ready to begin clinical trials of their enzyme. We also learned that, while they were going to start by treating four babies, they were also going to include three late-onset patients in their study. However, to be included in the study, a person could not be ventilator-dependent. This meant that it was too risky for me to have the surgery, because one of the biggest worries was that I would be unable to be weaned off of the ventilator afterwards. So we waited, and in June 1999, my mother and I moved to Rotterdam, the Netherlands, and I became the first late-onset AMD patient in the world to receive enzyme replacement therapy (ERT). For the next year, I received weekly infusions and had numerous tests done to see if the treatment was working. This was a very stressful time for the entire family, but it paid off. There were no drastic changes, but I started to feel better. However, my scoliosis was getting worse and worse, and it became imperative that I have surgery to correct it.

Before I talk about the surgery and what happened afterwards, I want to talk a little about my experiences in Holland and some of the incredible people that I met while living there. First, I have to say that while I believe that I owe my life to my parents, I also would not be alive today if not for the supreme dedication of the "Pompe Team" in Holland, especially the leaders of the team: Dr. Arnold Reuser and Dr. Ans van der Ploeg. These two doctors spent over a decade working to find a treatment for Acid Maltase Deficiency and didn't give up until they did. In addition to that, while my mom and I were all alone in a foreign country, they went far beyond what was expected to make us feel welcome and comfortable. Dr. Reuser even took my mother and me on several different outings to see castles and other famous landmarks that we would not otherwise have seen. For these reasons, I will always hold a special place in my heart for them, and I will always be grateful to them.

Another of the doctors on the Pompe Team was Hanneriecke van den Hout, a young doctor who was working on her Ph.D. For the first six months that we were living in Holland, it was only my mother and I (my sister joined us in December), so I did not have anyone else to talk to. Every week when I was in the hospital for my infusion, Hanneriecke would stop by and talk to me. They were never deep conversations; in fact they were usually about my family or what I had done during the week (which usually was not much). However, these conversations gave me something to look forward to and helped me not feel as homesick as I might have otherwise.

It was another incident, however, that will always stay with me and remind me of how good a person Hanneriecke is. As part of the clinical trial, I had to have several muscles biopsies (a muscle biopsy is a procedure where the surgeon makes an incision and then removes a piece of muscle for study) so that the doctors could see whether or not the treatment was helping to clear the glycogen from my muscles. However, because it was so dangerous for me to be given a general anesthesia, I had to undergo all of these biopsies under a local anesthesia. This meant that I was awake during them. The first biopsy was to occur before my first infusion and they were also going to install a port-a-cath (an implanted device that allows for easy access to a vein) for use during my infusions. I was very nervous because the only biopsy that I had ever had before was done under a general anesthesia. I guess Hanneriecke could see my anxiety, because she stayed with me throughout the entire procedure, holding my hand and talking to me to keep my mind off of what was going on around me. Her actions made the whole process bearable, and I will never forget them. If I had not had the ill fortune to have this disease, I never would have met these incredible, dedicated people.

When it became time for me to have the scoliosis surgery, my parents decided to return to the U.S. in order to have it done. So, after several months of negotiations, I finally got permission to receive the ERT treatments in the U.S. As soon as we were given the green light, we packed up and flew to Rochester, Minnesota where I was going to have the surgery. Once again, Bodie was invaluable in helping us to get things done. We arrived back in the States on June 27, 2000, and three days later I was wheeled into surgery to have the first of two surgeries to correct my severe scoliosis. By this time my scoliosis was at 100° , and I had been confined to a wheelchair for over a year. However, my surgeon, Dr. Shaughnessy, did a superb job. When the second surgery was over, my curvature was less than 35° . Although I spent the next three months on the rehabilitation floor of the hospital, and I was in a lot of pain, things were starting to look up. I had been able to wean myself off of the ventilator a few hours after the first surgery, and a mere 7 days after the second one, something that the doctors did not think I would be able to do. In addition to that, I had a new, and improved body. It may sound trite to be worried about how my body looks when I am dealing with a deadly disease, but as my scoliosis increased my self-confidence had decreased drastically. After the surgery, I started to get my confidence back, and I was much happier for it.

The next big step for me was returning home. I had not been home in a year and a half, and when I left, I was still walking. To say that I was nervous is an understatement. However, when I saw my entire family again, as well as assorted friends, my worries started to fade. I had been worried that everyone would treat me differently now that I was in a wheelchair, but they didn't. I have to say that one of the people that I was the most nervous about seeing was my cousin, Kellen. He and I are the same age, and we have always been close, despite the fact that he does not live in Texas. I saw him again for the first time about two years after I returned to San Antonio for good, in the summer of 2002. He put all of my fears aside when he just sat on the ground next to me, and we talked for the next two hours. Then, the following Thanksgiving when he came to town, without any words being passed between us, he would simply pick me up and put me in the car if we were going

somewhere, or push my wheelchair to whatever our destination was. His actions have made me feel more comfortable about my situation, something that I will always remember. In fact, as time has passed, I am less and less self-conscious about the fact that I am in a wheelchair, and about the things that I am unable to do. Yes, I am in a wheelchair, and I don't know if I will ever be able to walk again, but that is okay because at least I am alive, relatively healthy, and I have a great support system of family and friends.

This does not mean that things were or are perfect. I still had to fly up to the Mayo every week for infusions for six months until we were able to get permission to have the infusions here in San Antonio, in April 2001. Then a year after I was home for good, I had to deal with the fact that the form of treatment that I was on was being replaced by a new form, whether I liked it or not (for more information go to the AMDA website at http://www.amda-pompe.org). Then, I had to deal with the fact that just as I was starting to feel pretty good, the change in treatment had negative effects on my progress. Despite all of these changes, I had started college, and so I had to deal with going to school again for the first time in over five years. But I have dealt with these things, and, now that my treatment dosage has been adjusted, I am starting to feel better, again.

Having Acid Maltase Deficiency and being one of the first patients to receive treatment for it has changed my life in numerous ways. It has made me very aware of how lucky I am to be one of the few patients that are receiving treatment. In fact, this is something that I have had to struggle with because when I see, email, or speak to the patients that aren't receiving treatment, I feel extremely guilty that I am receiving it while they are not. It is especially hard when I hear about the babies and young children that have died because they did not receive treatment in time. But, I believe that I am a stronger person for the experiences that I have had. I had to grow up fast, but I grew into a person who can deal with almost anything that life throws at her. I have also learned a great deal about myself. The most important thing that I have learned, and that I am very adamant about, is that my disease does not define who I am. I am not only a person with Acid Maltase Deficiency. Who I am is a person who happens to have AMD, and whose experiences for the last nine years have been shaped, in part, by this fact. But, I refuse to limit my view of myself, or to let anyone else limit their view of me, to only this single fact. I am also a twenty-year-old girl, who is the oldest daughter of Randall and Marylyn House, the older sister of Andrea and Randy. I am an English and History major, I enjoy reading and spending time with my friends and family, and I want to be an editor/publisher when I graduate. These are just a few of the things that define me, and as I continue to grow as a person, the list of things that define me will grow as well. But I will *always* refuse to be defined only by my disease.