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COMMENTARY

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Thalassemia: Yesterday, Today, Tomorrow

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1 | YESTERDAY

It was the early 70s, I was 2½ years of age when my pediatrician told my parents I had Mediterranean Anemia. I grew up in Astoria, a Greek enclave in Queens and that's how my disorder was called. My parents, immigrants from Cyprus, had limited command of the English language, and the news that their baby daughter had a fatal disease tore them up. My parents did not know they were carriers of the genetic disorder thalassemia. They did know about the disorder because in Cyprus it was known as the "STIGMA" (a mark of disgrace or infamy). In real life, the word took on a more troubling definition as many new parents considered the birth of a child with thalassemia as a family shame. My parents took the very opposite position: They were brave and defiant; everyone knew that Maria had thalassemia. My parents also instilled confidence in me at a very young age and they taught me to never be embarrassed to speak about my disorder and never let anyone make me feel ashamed.

It wasn't easy being a child and feeling like a human pincushion. I had many moments of discomfort and pain from the constant needles and treatment. Each time, I knew my parents felt that pain 1 million times over. The veins of a thalassemic child are small, slippery little suckers!! Scar tissues started forming because of the multiple sticks. Often, I was stuck from half a dozen to a dozen times. Sometimes, the nurses had to transfuse me through a vein in my legs because the veins in my arms were damaged.

By the time I reached nine years old, my spleen weighed almost 6 lbs and had to be removed. Today, this procedure is not as common because our treatment has improved. The ability to fight infections, viruses, and bacteria without a spleen is dramatically reduced. The hospital became my second home, and I fought raging fevers up to 105 degrees. During one scary episode, the IV antibiotic drip was not working, so the nurses submerged me in a bathtub filled with ice. It felt like hours had passed when I woke up lethargic, shivering, and teeth chattering. What I most remember is seeing my mother sitting on a steel-folding chair holding the Bible. I asked my mom twice "Why is this happening to me?", and each time she gave me a great answer. At the third time, she got off the chair, set the Bible down and said: "Look at me, there is no cure for your disorder, so you decide how you will live with it for the rest of your life." My decision was to live a happy life. How does one live a happy life with a chronic disorder? You become friends with it. I embraced everything about thalassemia.

That same year my ferritin was at a level over 2500, and I started an iron chelator, deferoxamine 5-6 nights a week to remove the toxic iron from my liver and other organs. Back then, I had to stay in the hospital for one week to learn how to mix the medicine and how to insert the butterfly needle in my tummy's or in our legs, if I ran out of spots on my tummy. I developed abscesses and cyst like bumps as the needle was inside of me for 10 h. The first models of the pump were huge and impossible to hide. But it got me through a decade of chelation from the 70s through the 80s. We knew the consequences—if we did not chelate we would eventually die of organ failure. Most patients in my generation started chelation too late or gave up because it was too difficult. When my father picked me up from the hospital at the end of chelation training, he looked at me and said, "This is your responsibility." To this day, there is no going against a strict Greek father. He would have no problem smacking me across the face if I didn't chelate. I also took it in a very different way; I felt empowered to be responsible for my health. Until then I wasn't given many responsibilities—just do well in school and keep my room clean. Chelation was a whole different level of responsibility, and I loved it.

At 11 years old my gallbladder and about 30 gallstones had to be removed. I gave up eating ALL junk food; sodas, chips, fast food, bad chocolate. . ..I cleaned my diet from trans fats and everything not healthy. As I entered the early teen years, I started to feel self-conscious of my body. In those years of surgery, they still stapled the skin together rather than using the more advanced scar-less technology of stitches and now laser. The thought of these scars lingered and caused self-esteem issues. There was never enough support in the clinic for doctors to speak to patients about these issues. Asking the question "And how are things at school?" is not a supportive enough conversation starter to get the young patients to open up and talk about what is real—the scars that no one can see, the internal scars

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that hurt more than thalassemia. The hospital visits were about getting transfused but luckily, I was being transfused in a clinic with 30 other thalassemia patients and we formed friendships, tight bonds and together made it through the tough adolescent stages. When it gets ROUGH, a thalassemia patient knows how to get TOUGH. We were a close bunch and laughed during the 8-10 h transfusion days; we made the very best out of the very worst. Other issues came into play with teenage growth and a delayed puberty due to the iron overload in our pituitary glands. I speak for many of us who struggled with the implications of fitting because of the delay in developing breasts and in getting our menstrual cycle, with some girls not getting it at all. I became very blessed when all that set in beautifully at age 18.

You expect teenage girls to be mean, but you don't expect harsh statements from your medical providers. As a child who grew up in the hospital, I repeatedly heard doctors on rounds tell young residents about a host of negative outcomes expected if you had thalassemia. They taught the young residents that I would never be a mother. I would not live a long life. I would not make it into college. Here's one thing I never heard. . .MY NAME because they never diagnosed me, Maria Hadjidemetriou. They just repeated the textbook diagnosis on Thalassemia, from the 50s when they went to school. Doctors who made their rounds with younger resident doctors called me by my Medical Record Number—a number I still have memorized. I would call my mother at night crying, telling her the things they said about me. My mother"s steady and wise response was "Don't cry, Maria. Remember in one ear, out the other. They are not God."

As the years went on, I started to THRIVE. I entered college, spent six months in Florence, Italy on a scholarship for International Marketing, traveled to 20 countries, graduated college and went to work in my field. I thought I was unstoppable, but one thing did stop me—the inability to gain Health Insurance. Like all people with pre-existing conditions, we now had to deal with this demon. The need to change this insane barrier to my future should have compelled me to be a better advocate for myself and others. To make the situation more infuriating, my hematologist still treated me like a diseased child. I realize now that the real disease was the mindset of my hematologist, not thalassemia. I became afraid. I didn't look for a full-time job with a design studio, I freelanced, here, there, everywhere making very little money. Nothing was solid. I cried so much because I wasn't excelling in my career. Here I was, a dynamic woman starting an exciting career, diminished because of my doctor's limited point of view.

During my twenties, I started learning more about my disorder through the only non-profit foundation in America. Founded in 1954 by the Ficarra family and other young parents, the Cooley's Anemia Foundation had grown to be a major influencer and advocate for patients. I started reading research papers and attended my first international conference in Bangkok, Thailand organized by the Thalassemia International Federation headquartered in Cyprus. Meeting patients, doctors, clinicians from all over the world was life changing. The international conferences offered a very rare opportunity for patients to interact and build relationships with doctors—a different breed of doctors. This meeting was the first time I heard about a new drug, deferiprone. It was the first time I met a thalassemic woman who had given birth. It was the first time meeting thalassemics who held full-time professional jobs that they loved. I became determined to achieve all of these things for myself.

Around this time in my life, I was also blessed to meet a clinical nutritionist who taught me the most vital lesson. Her words were "You are the most important partner in your health. . .YOU." I started taking supplements, vitamins, and antioxidants, a regimen I still follow today and which has powered me through the past few decades even with Osteoporosis. All the years of chelation removing the toxic iron had also stripped my bones of calcium, magnesium, and zinc. I did what came naturally to me: I got empowered to exercise despite fractured ankles and acute pain. Five times a week, I am weight training or taking a Yoga or Pilates class because I am responsible of keeping this body strong and patients tend to forget this responsibility.

In 2001, I felt something wrong with my heart: as a resident of downtown Manhattan during the 9/11 attack, I thought my anxiety and symptoms might have been related to that trauma. I told my doctor that my heart hurt, so they ran several cardiac studies all negative. Still, I knew this could be iron overload in my heart and without tools to directly measure the effect of iron in the heart available in the US at that time, I made my way back to Europe for an MRI test called T2*. The test showed that my heart was loaded with iron. I knew I needed to stay in Cyprus because I needed access to deferiprone, which was not available in the US. In Cyprus, I began using deferiprone and 14 months later my heart became healthy and strong. The experience of being treated with thalassemia in Cyprus was wonderful. I was treated as a person, not a patient and empathy was paramount to the team during each hospital visit. I met many thalassemia moms including one who had four children. I could work and thrive without health insurance and career stress. I stayed in Cyprus for three years and returned to NYC in 2005 stronger than ever. Deferiprone was still not approved in the US, but my family continued to send me supplies from Cyprus. I was doing very well, in love for the first time and gave birth to my wonderful daughter Julia in May 2008.

2 | THE FDA EXPERIENCE

In 2011, after working with the Cooley's Anemia Foundation over the past six years to repeatedly make the case to the FDA for the approval of deferiprone I was invited to testify at the Advisory Committee hearing. Over 50 people were present, and other patients and world-respected hematologists and radiologists testified. In my bones, I knew that the approval would happen that day. Many controversies, egos, and ignorance held back this drug. During our long wait, I lost a lot of friends to cardiac failure and inability to stay on or ineffectiveness of other chelators. My dearest friend passed just after the approval, and I still hear her encouraging me and others to keep up with our chelation and to carry on as an advocate for others.

3 | TODAY

My daughter will be nine years old in May. We continue to live, love, laugh each day in our cozy Lower Manhattan apartment.

My passion is building awareness about thalassemia. The recent release of a documentary I wrote and produced and which featured my story, Thalassemia: Life Without Boundaries, is a major step in my continued quest to bring awareness to this disorder. The title expresses how I live my life and my aspirations for others.

My current chelation is the combination of daily deferiprone and deferoxamine four times a week. My cardiac iron levels are excellent, but there is moderate iron overload in the liver. I recently underwent an MRI for my pancreas and my pituitary gland, and they found evidence of severe iron overload. My bones continue to deteriorate, and my Dexa score is as low as −4, but I am not giving in to the pain.

4 | TOMORROW

I am very hopeful that there will be a cure in my lifetime, maybe the cure is not a perfect one for adult thalassemics, but I will be elated to see this for the younger thalassemia patients. I do not want them to have to endure any pain into their adult life. There are promising research studies with new drugs which may lessen transfusion requirements and the resulting iron burden. Successes have been reported with gene therapy. I continue to live my life with a positive attitude and teach my daughter to do the same. This legacy began with my mother's courage and insistence that I decide on how to live. We have a choice, no matter what God gives us we have a choice. My view is with the right attitude; we will make the right choices.

URL: http://bit.ly/ThalassemiaLifeWithoutBoundaries

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