



Newborn Screening
Saves Babies
One Foot
at a Time

Kristina

THIS IS A PERSONAL STORY OF GROWING UP WITH
CARNITINE PALMITOYLTRANSFERASE TYPE II DEFICIENCY (CPT II).

For as long as I can remember I had bouts of intense muscle pain. As early as age 7, I helped with my sister's paper route, and some days I was fine, and others I'd reach a point when I could no longer walk. I'd stop, start crying and wait for my parents to realize I'd been gone too long. My dad would come searching for me and carry me home where I'd sit for hours, my legs feeling as if they were on fire.

Later I tried sports. I remember the horrid pain of gymnastics, ice skating lessons where my legs felt they were freezing up, and swim practices when I swore my arms and legs couldn't move one more foot. But throughout my early and middle childhood I continued. My pediatrician said the pains were probably just growing pains

As I entered middle school and began swimming more seriously, the bouts of pain came less often, but when they came, they were much stronger. I saw a sports medicine doctor who tested me for all sorts of things, without any positive results. I had several major episodes during which I couldn't walk and it hurt to talk. I was diagnosed with "heat exhaustion" and "stress." Finally at age 15, after many tests and doctors' appointments, my doctors and parents suggested that perhaps I was making it all up. Over the next two years I had several major episodes—one during a marching band parade performance and another during a week of double-sessions of swim practice. During the episodes the pain was so intense that I could not walk for days, needing help to get to the bathroom, all the while, thinking it was all in my head. I went to bed crying many nights, alternating between thinking that my doctors and family were wrong, and then becoming thoroughly convinced they were right and that I was crazy.

Finally, the day that ended up making all the difference happened. I woke up one September morning, psyched about the big swim meet that day—we were to compete against our biggest rivals. I woke up and didn't feel quite right. I had a fever but went to school anyway (after all I couldn't swim in the meet if I wasn't at school that day). I made it through the first hour of class but suddenly the muscle pain began. I struggled to walk down the hall to second period, but it got worse. Within moments my whole body was in pain. I made it through the end of class and then went to the office to call home. By the time my ride arrived I couldn't walk. My mother and her friend carried me into my room where I lay for an hour. The pain just got worse.

Finally my mother had had enough. She got me into the car, drove me to the doctor's office and made a scene—a very important scene—she said she wasn't leaving until a doctor saw me. My pediatrician was gone that day, so I saw a new one. I told him how every muscle hurt—it hurt to walk, talk, breathe, bend my fingers and even blink my eyes. Looking over my chart, he decided to give me Valium (after all, the previous doctor had concluded it was psychological) which didn't affect the pain at all. Importantly, he also asked me to pro-

vide a urine sample. I looked at it and saw that it was dark brown and thick but had no idea what it meant. While I waited for hours, I hit an all-time low. I asked my mother to kill me. The pain was just too much and I couldn't take it anymore. I thought I was surely going to die, so why not speed up the process?

That night the doctor had me go home. I was awake all night trying to be still, trying to breathe small breaths. First thing in the morning we got a call to go to the hospital. My CPK was over 96,000 (it should be under 200). And with that, the testing began. Seven months and many tests later, we had a diagnosis—Carnitine Palmitoyltransferase II Deficiency. I'll never forget the day my doctor called to tell me the diagnosis was in—I cried. I was so overjoyed to discover that there was a name for the way I had been feeling and that it wasn't a figment of my imagination.

With the assistance of a diagnosis, I've become much more able to deal with my illness. I know the events and lifestyle choices that trigger it. I know how to respond, and I'm getting pretty good at knowing when I need to see a doctor vs. when I can treat myself at home. While I have had to give up some activities, I continue to go to the gym and stay active, traveling overseas and attending a full-time Ph.D program. With a healthy diet, regular exercise and sleep, I am able to live a mostly normal life. In fact, most people I know have no idea I have a health problem until I tell them. From time to time the inevitable episode of muscle pain happens, but I deal with it and get back to life quickly.

In the last few years I've been a part of pioneering treatment and research studies and have met a great community of individuals who live with CPT II. Unfortunately, each time I meet a new "CPT-er", similar elements of our stories emerge—years of unexplainable pain, misdiagnosis and frustration. I am optimistic however, that future generations of "CPT-ers" will be diagnosed earlier as medical professionals learn about this disease and as more children are screened at birth. This will allow them to live full lives with only a few modifications and to avoid the physical and emotional pain of years of misdiagnosis. For now I'm happy knowing what's responsible for the pain I feel and, more importantly, for knowing how to deal with the pain when it comes up.

Kristina

Written August, 2006