

PKU: Kristi - An Adult Looks Back on Her Experience



My mom received the phone call one Saturday morning shortly after my birth, and her heart sank. “Your daughter’s PKU test came back positive. PKU causes mental retardation*. They’re trying a new special diet that will help improve her IQ by a few points. Come in on Monday,” were the pediatrician’s words on the other end of the line. As she looked at her perfect baby with 10 fingers and 10 toes sleeping like an angel, she thought, “Kristi can’t be mentally retarded!”

Mom and Dad hit the University of Arizona medical library that weekend and realized that the most up-to-date books on PKU treatment spoke of a special low protein diet that was not “new” but had proven to allow children with PKU to lead normal lives and they were starting to encourage “diet for life.” Some patients who had been diagnosed with PKU and treated as infants

were beginning to hit adulthood and function normally. By Monday’s appointment, my parents knew more about PKU treatment than my pediatrician.

When I was younger, my parents struggled with the stress of making sure I ate foods that were on my diet, drank my “special milk” multiple times every day (which provides me with the nutrients my body doesn’t get due to limited diet), had appropriate snacks at birthday parties and school events, struggled with the finances that insurance didn’t want to cover, and presented the PKU way of life to me as “no big deal.” PKU was a part of my life, but it did not define who I was or what I could do. I am told that I got in a power struggle with a new teacher at daycare when I was about three because I refused to eat that day’s snack of cheese and crackers. I was a quiet, shy, ready-to-please child so the tears and refusal surely shocked the teacher. After some time, a seasoned teacher walked by and figured out what was going on. I knew I was not allowed to eat anything unless it was packed by my mom or dad. Kudos to my parents for teaching me that rule so young!

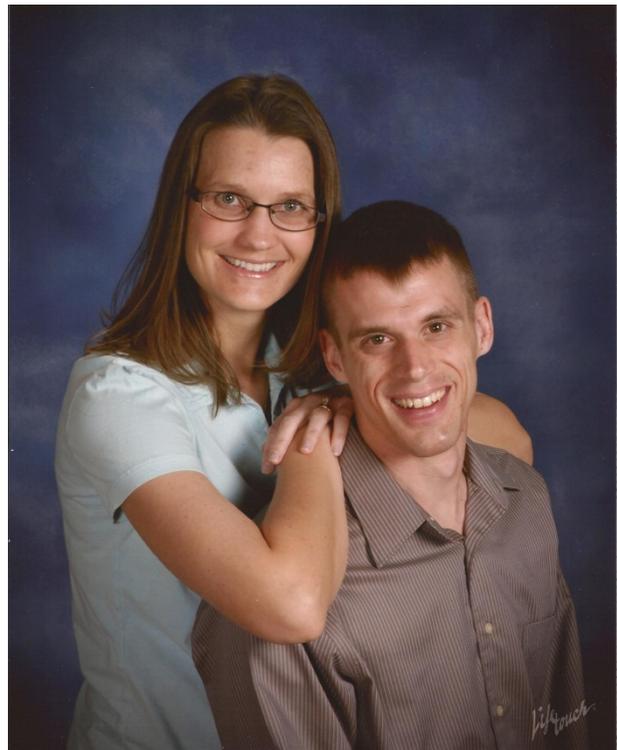
As mentioned before, I was quiet and shy. This presented a challenge when I decided I wanted to “blend in” and eat from the hot lunch line at school. Every child going through the hot lunch line was required to get the entrée, a certain number of sides, and milk. Supposedly you could buy certain items, but this never worked out for me. The lunch monitors gave me a hard time, I felt embarrassed, and my hot lunch days were over. My mom offered to talk to the school but I refused--I did not want the extra attention. In middle school, I discovered the cafeteria had French fries or tater tots every day and I was able to buy “hot lunch” items without the hassle I had before. I learned that pickles, ketchup, and low protein bread made a great sandwich, though it got weird looks from my lunch table. I’d explain, “It’s like eating a hamburger without the burger!” People would shrug it off and continue with their lunch.

PKU wasn't so bad after all! In high school, I decided salads were a better option and ate a salad, chips, and a tootsie roll for lunch every day.

It turns out the books my parents had read were right. By high school, I was in honors classes and was NOT mentally retarded. I decided that I wanted to go to college. My parents made a deal with me that if I got my college paid for, they would buy me a car. I loved soccer, but I knew that my chances of going to college on a soccer scholarship were slim. I figured out that if I was in honors classes, I could probably score an academic scholarship. Fast forward a few years and I graduated in the top 2% of my senior class, earned an academic scholarship to any in-state university, received the car my parents had promised, and proved to myself and many others that PKU children could be good at math, science, and life in general.

Today I have two bachelor's degrees: my first one in public health with a minor in molecular and cellular biology, and my second degree in nursing. I continue to follow my low protein diet, check my blood levels twice a month, and drink my "special milk" every day. I have just completed my term as founding President of the Arizona Network for PKU and Allied Disorders (ANPAD). I am married to a wonderful man who learned about PKU when he met me, but if you spoke with him, you would think he's lived with PKU his entire life. We are active in our church, volunteer with our church's high school youth group, play soccer together in a co-ed recreational league, and are preparing to become foster parents to children in need. I work as a nurse at a pediatrician's clinic, where ironically, our newborns are screened for PKU and other disorders.

I understand that people don't want to take their child in for the blood test and cause pain, but I am able to speak first-hand of why we do these tests. Newborn screening truly does save lives - it did mine!



***Editor's note:** In the early 1980s terminology like this was common and is included in this story for emphasis and true-to-life storytelling. The medical community no longer uses these terms. We apologize if readers find this language offensive.