

I want to introduce you to my eight-year-old daughter, Claire. At first glance Claire appears to be a normal 2nd grader. She loves playing soccer, gymnastics, and playing with her brother and sister. She's silly and funny and just a joy. What you don't see is that she has an extremely rare metabolic disorder called Tyrosinemia Type 1. We were very lucky that her disorder was discovered through newborn screening. Louisiana expanded their testing two months before she was born. Without this expanded screening, her outcome could have been fatal.

So, what is Tyrosinemia? Basically, Claire's body does not properly break down the amino acid, tyrosine, which is a building block of most proteins. Since the tyrosine cannot be broken down, it becomes toxic to her body. This can cause liver and kidney failure, softening and weakening of the bones, problems affecting the nervous system, and increased risk of liver cancer. The medically necessary treatment for Tyrosinemia Type 1 is a low protein diet, a metabolic formula, and medication. Specifically formulated low protein foods are essential for necessary calories and for ensuring an adequate quantity of food, as well as, providing variety and increasing quality of life. The metabolic formula provides the necessary protein, calories, and other nutrients in a form free of tyrosine. This is a lifelong disorder. There is no cure.

What does it mean to have a "low protein" diet? The low protein diet has two components: food and formula. For example, Claire can only have 10 grams of protein per day. My cell phone is about the size and weight of a 5-ounce piece of chicken. This would be 38 grams of protein ... 4 days' worth of food for Claire. This illustrates why it is so important that patients with metabolic disorders have access to low protein foods. Unfortunately, the cost of low protein foods can be very expensive and not uniformly reimbursed by health insurance, resulting in a large financial burden on a family. For example, a bag of low protein pasta is \$16, and a bag of veggie meatballs can run around \$24. And shipping these special foods can be very costly as well since you can't pick them up from your neighborhood grocery store. Then there's the metabolic formula. A case of 6 cans is about \$500 and we use about 1 case per month...\$6,000 per year.

Many states do not assist in formula coverage and do not require insurance to cover medically necessary foods and formula. Some states only cover certain metabolic disorders like PKU, while other states cut off coverage to males over age 21 years. This is dangerous, inconsistent, and unacceptable. This is a diet for life. Claire depends on medical nutrition to keep her healthy and growing.