picture page 50

What will Briana Do?
A Family Story about PKU

Hi. My name is Amanda and I am 15 years old. I have a little sister Briana, 13 years old, who has a genetic disorder called PKU. This disorder means she has a really strict diet of only fruits and vegetables and has to drink a special formula three times a day. She isn't allowed to eat very much either and so she is hungry a lot. If she doesn't stay on this diet she becomes mentally retarded. I remember when she was little and both she and my mom would cry because she was hungry and too little to understand why she couldn't eat anything else for the day.

Everything we do as a family is affected by Briana's diet. When we eat out we always have to plan ahead so we can make sure that they have something she can eat. At home we always cook plain vegetables so Bri can eat something the rest of us eat. A lot of times when Bri goes to a party she can't eat anything that is served so we make or buy her special low-protein foods that are very expensive. It is amazing how many times people eat or plan activities around eating. You don't notice until you have someone in your life that has to have a special diet.

In three years I will be going to college and in five years Briana will too. I am wondering how she is going to be able to manage her diet and deal with living on her own. The special foods she has to eat are expensive and her formula is *really* expensive. The State will help her with her formula until she is 21. But, what happens after that? My mom says it costs about $550 a month for her formula and just one-pound of low-protein spaghetti noodles cost $6.00. I don't know how you pay rent and all of your other living expenses *and* pay for formula and low-protein foods at the age of 21. It is pretty scary for all of us to think about that right now. I love my sister and even though I think she is annoying sometimes, I want her to be healthy. I am worried about what is going to happen to her in the future. I hope she will be able to be okay.

Phenylketonuria is an inherited disorder of amino acid metabolism. Individuals with PKU are unable to metabolize the amino acid phenylalanine (PHE) in a normal way. This defect in protein metabolism causes PHE to build up in the blood and results in brain damage. All newborns are screened at birth for PKU. To prevent mental retardation, a special diet low in PHE is required and must be initiated within the first month of life. For optimal neurological health, this diet must be life-long. Affected women of childbearing age must be on the diet prior to conception and during pregnancy to prevent mental retardation of the fetus. In adults who discontinue treatment, complaints of agoraphobia, depression and anxiety occur, which subside with treatment. Without adequate treatment, behavioral disorders, aggression, self-abuse, hyperactivity, impaired thinking, altered perception and memory, make it difficult for the individual with PKU to perform the daily activities of life. The cost for formula and low-protein foods are high. All children in Oklahoma, under age 21, can obtain the prescribed formula free. Women with PKU who desire to become pregnant also can obtain free formula. However, there is no subsidy for the low-protein foods. Serving sizes of regular food are limited for an individual with PKU. For example, a comparison of serving sizes between regular food and low protein would be a choice of one regular saltine cracker or twenty-five low-protein crackers. If you were hungry, which option would you rather have? When low-protein foods are not available, complaints of hunger are frequent resulting in the person being tempted to eat regular foods or to go off diet. The cost for the medical treatment for PKU can be a burden to a family and is often not covered by insurance.