Glut1 Deficiency Syndrome: A Little-Understood Disability

By Emma Hemness, CELA

Before representing the Special Needs Alliance (SNA) at the Glut1 Deficiency Syndrome Foundation's annual conference in Orlando, I knew very little about the disease being discussed. Unfortunately, my lack of awareness wasn't unusual. According to the National Institutes of Health, since first identified in 1991, only a few hundred cases of Glut1 Deficiency Syndrome have been reported worldwide, although it's believed that thousands of unidentified or misdiagnosed cases may exist in the U.S. alone.

Glut1 Deficiency is a genetic disorder that prevents the proper transfer of glucose to the brain. Symptoms vary greatly and may include seizures, motor problems, developmental delays, learning disabilities and more. It's a spectrum disease, with symptoms that may range from minor to severe, and be continuous or intermittent. Physicians themselves often harbor misconceptions about the condition, focusing on a narrow set of symptoms manifested during childhood, when the condition is lifelong. For these reasons, as well as the fact that there's seldom a family history to point the way, misdiagnosis is common, and it may share symptoms with other diagnosed conditions, including cerebral palsy, epilepsy and ADHD.

It's not unusual for families to visit half a dozen specialists over the course of as many years before reaching an understanding of their child's health difficulties. Yet early treatment is important to safeguard brain growth during critical periods of development. Available diagnostic tools are part of the problem. The spinal tap is often the first step of evaluation, a procedure that can be painful and involves a certain amount of risk. Genetic assessment is another route, but it's not available everywhere, can be expensive and may sometimes result in a false negative.

While there's no cure for Glut1 Deficiency, most individuals respond well to the *ketogenic diet*, a high-fat, low-protein, low-carbohydrate regimen that replaces glucose with fat as an energy source. The diet should be tailored to each individual and monitored by a medical professional. Side effects exist, and some find the dietary restrictions difficult to maintain. More research is needed concerning the diet's long-term use.

Challenges of a Rare Condition

Lack of awareness and widespread misconceptions mean that families dealing with Glut1 Deficiency may face difficulty obtaining the supports to which they are legally entitled. They

may find that they don't neatly fit into the disability definitions adhered to by state and federal benefits programs. They may also have problems obtaining the special education services needed by their children.

Since this is a spectrum disorder, not all families will need to do special financial planning for their child. But those dealing with Glut1 Deficiency's more severe symptoms should certainly think about long-term security and quality of life. Establishing a special needs trust to which friends and relatives can contribute should be considered as a means of protecting a child's eligibility for means-tested benefits, such as Medicaid and SSI (Supplemental Security Income), which may be needed in the future. Parents should not delay in consulting a special needs attorney to put in place their own estate planning documents to help protect their children.

Organizations such as the Glut1 Deficiency Syndrome Foundation are important sources of support and information for families that often experience a profound sense of isolation. Meeting parents and grandparents at this summer's conference was an eye-opening education for me. I encourage others to learn about and spread the word concerning this little-understood disorder.
