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Pediatric Neurotransmitter Diseases

by Anne Berleman Kearney

You probably do not know much about pediatric neurotransmitter diseases (PNDs); not many do. But it is possible that many undiagnosed disorders in children are pediatric neurotransmitter diseases.

The term "Neurotransmitter Diseases" is an umbrella term for rare genetic disorders that affect the functioning of central nervous system neurotransmitters and the metabolism. Neurotransmitters are chemicals that permit the passage of signals within the brain. Many neurotransmitters within the body work together to regulate motor coordination, behavior, temperature, pain mechanisms, and blood flow. A commonly known neurotransmitter is dopamine, a deficiency of which is associated with Parkinson's Disease.

Neurotransmitter Function and Disease

The processes by which our bodies make, use, and break down neurotransmitters is extremely complicated. When there is a disruption in these processes, problems or abnormalities can occur in essential body functions. We see manifestations of these problems in the various neurotransmitter diseases. Common signs of these diseases include hypotonia (low muscle tone), rigidity, movement disorders (tremors or dystonia), oculogyric crises (abnormal eye movements), temperature instability, profuse sweating, absence of speech, and developmental delay.

We began our search for answers after our middle son, Stephen, began to show some of these symptoms around his first birthday. Stephen then--and now--has significant hypotonia. He cannot speak. He also suffers from temperature instability. Stephen has been diagnosed with a pediatric neurotransmitter disease, Succinic Semialdehyde Dehydrogenase (SSADH) Deficiency.

SSADH is one of the disorders that falls within the pediatric neurotransmitter diseases umbrella term. The other disorders are:

- Tyrosine Hydroxylase Deficiency (TH)
- Aromatic L-Amino Acid Decarboxylase Deficiency (AADC)
- Guanosine Triphosphate Cyclohydrolase I Deficiency (GTPCH), in multiple forms
- Sepiapterin Reductase Deficiency (SR)

The means of diagnosing pediatric neurotransmitter diseases varies from the invasive testing of spinal fluid for some disorders, urine testing for others, and a mass spectrometric blood analysis for still others. Stephen was diagnosed through urine testing.

Treatment

Treatment for some of these diseases is drug therapy, while behavioral and psychosocial interventions are important for other of the diseases. For example, for children like Stephen with SSADH Deficiency, physical and occupational therapy, including swimming and horseback riding, are beneficial in conjunction with their special education classes and speech therapy.

My son, Stephen, has made slow but steady progress in following such a regime. An intense schedule of physical and occupational therapy, including swimming and hippotherapy, has helped him with balance and strength. Perhaps the most important interventions for Stephen have come through sensory integration therapy. Not only has sensory integration therapy helped with strength, it has given Stephen a better sense of how his body moves in space. Happily, just this past month, he took his first independent steps at nine years old. In addition, we continue with an intensive speech therapy program to work on vocalization, and we have added a simple wooden letter board, along with a more sophisticated augmentative communication device, as alternate means of communication.

Support for Children with PNDs

My family has been lucky to be part of a support network that exists for families of children with PNDs. The PND Association (www.pndassoc.org) was formed in 1998 by a group of parents whose children had the hallmarks of neurotransmitter diseases or had been diagnosed with neurotransmitter diseases. Today, they are joined by a medical and scientific board comprised of leading doctors and researchers in the fields of neurology, genetics, and metabolics.

The PND Association has been a catalyst for raising awareness of these rare diseases within the medical profession and the public, increasing diagnosis (in part, through the funding of diagnostic equipment), and promoting treatment. Of course, the overriding

goal of the PND Association--and for us--is to find cures for these pediatric neurotransmitter diseases.

The PND Association receives about four inquiries per week from families or professionals for testing and disease information. Over the years, there has been a distinct rise in the detection of pediatric neurotransmitter diseases. And, for some of these diseases, that detection leads to treatment that can work miracles—big and small. Our hope is for more of these miracles.