

Report Of a Case of Double Vision - Sept. 2008

By Rick Rader, MD, Editor-in-Chief

Exceptional Parent Magazine

Sep 1, 2008 - 1:35:29 PM

Last week in Washington, I saw double.

It was more like a split screen than double vision. While it was dramatic and stopped me in my tracks, it didn't cause me to rush to my ophthalmologist or start to think of a name for my future guide dog. But sure enough last week in Washington, I saw double.

I was in Washington to attend the Pediatric Neurotransmitter Disease Symposium tagged "The Medical Management of Pediatric Neurotransmitter Disorders: A Multidisciplinary Approach." Not to be a namedropper, but the program had quite the pedigree, being presented by The National Institute of Neurological Disease and Stroke, The Office of Rare Diseases, The Johns Hopkins University School of Medicine, and the Pediatric Neurotransmitter Disease Association.

Neurotransmitter diseases is an umbrella term for the myriad of disorders that play havoc with the array of substances (neurotransmitters) that are released at the junction of one neuron and expedite the expected reaction on another neuron that eventually commands, encourages, or leverages an organ to do its thing. From here, we would start to trigger your yawn response unless you had the stamina to wade through the chemistry of monoamine degradation, dopamine, norepinephrine, and serotonin deficiency. Throw in some fundamentals of pterin-4a-carbinolamine dehydrates deficiency OMIM 264 070, and you quickly get the point that this is not a set of diseases that you would read about in a Reader's Digest, perhaps with an article title, "I Am Joe's Liver," although the liver is one of the culprits in this family of diseases.

Several years ago there were no "pediatric neurotransmitter diseases." That's not to say that these devastating disorders recently arrived from some alien life form. They have always been there, raging havoc, halting dreams, and changing plans. They're disorders that roll off the tongue like succinic semialdehyde dehydrogenase deficiency, guanosine triphosphate cyclohydrolase I deficiency, and aromatic L-amino acid decarboxylase deficiency. They roll off the tongue (cough cough), depending on what language your tongue is tied to.

Parents of children challenged with these disorders (parents....surprise!) who attended the conference became a family as they have always done. They were trading the names of researchers, physician champions, sources for non- FDA approved therapeutics, regimens and, of course the stock and trade of all exceptional parents, trading stories. There was a core of parents that finally decided to organize and funnel their energies. Carolyn and Brad Hoffman (parents of Sam who has SSADH) and Nancy Speller (mother of JJ, who, before he died, touched the hearts of everyone at EP) realized that no one was going to rally behind rare diseases that required two days of practice before you could pronounce the first three words of these complex conditions. They realized that there was strength in numbers (not that these conditions didn't already have enough strength on their own), and they mobilized to formulate and anoint a new class of conditions called "Pediatric Neurotransmitter Diseases." Those early meetings appeared to be like a sketch from Saturday Night Live. "Can we simply just give these disorders a new term?" "Don't we have to ask someone if we can do this?" "Who do we tell first?" They decided they could, and they did. Today, the Pediatric Neurotransmitter Disease Association (PND) is a force to reckon with. They gathered a world-class medical advisory board, raised money to fund new research projects, and established fellowships in pediatric neurotransmitter diseases. And the name stuck.

These are full tilt boogey disorders—disorders that impact on virtually every aspect of a child's development.

One of the sheer delights of being at a parent generated symposium like the PND Association is that in addition to the cadre of world class clinicians, researchers, and scientists, who not only lecture but are available to meet with the parents (at any time for any question) there are “the kids.” It's a true family affair, with not only the PND kids, but brothers, sisters, grandparents, aunts, uncles, and friends participating. Caring for a child with PND is a team sport and seeing the PND team (parents, clinicians, researchers, and supporters) made that clear.

Sometime on the second day of lectures (that gave me sweaty palms that I remember from my med school biochemistry lecture days), a world-class researcher was going over the biochemical pathways that were instigators in one of the rare forms of a rare neurotransmitter disease. The screen was wall-to-wall cave drawings showing where ALDH7A1-antiquitin-alpha amino adipic semialdehyde dehydrogenase has bowed out of the cadence to start to degrade the neurotransmitters. With a smooth gesture, he reached inside his sports jacket and pulled out a red laser pointer. The red dot began a circular dance on the screen, demonstrating where “ground zero” is in the pathway. It was dramatic, concrete, and lukewarm. It made as much sense to me as a game board from the Maldives; my grasp of biochemistry did not allow me to process what that meant to a child that was hosting the error or what it meant to their brothers, sisters, parents, and grandparents. And then it happened, my double vision. Out of the corner of my eye while still engaged in the swirling red laser, I saw Sam, or was it Ryan or maybe Swathi, being held by their mothers, and both overlapping images persisted. My double vision: the dancing red dot against the backdrop of a loving parent holding their precious child, deficient and lazy neurotransmitters to boot. The parents of PND kids are models for every exceptional parent. Parents who decided they could indeed, declare and announce a new class of diseases and start working towards a cure.

Yeah, last week in Washington, I saw double.