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EDITORIAL

International Symposium on Pediatric Neurotransmitter Diseases

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The second International Symposium on Pediatric Neurotransmitter Diseases (PNDs), entitled "Medical Management of Pediatric Neurotransmitter Diseases: A Multidisciplinary Approach", was convened on July 18–19, 2008, in Washington, DC, USA. The meeting was sponsored conjointly by the National Institute of Neurological Disease and Stroke (NINDS), the Office of Rare Diseases (ORD), the Johns Hopkins School of Medicine and the PND Association. This second symposium followed almost six years to the day from the first International Symposium (May 18–19, 2002), the proceedings of which were presented in a supplement to *Annals of Neurology* (Volume 54 Issue S6, pages S1–S109 (2003)).

There are currently five rare, inherited disorders that affect central neurotransmission and are housed under the umbrella of the PND Association. These are: succinic semialdehyde dehydrogenase (SSADH) deficiency (or γ-hydroxybutyric aciduria), tyrosine hydroxylase (TH) deficiency, aromatic L-amino acid decarboxylase (AADC) deficiency, guanosine triphosphate cyclohydrolase I deficiency (GTP cyclohydrolase, or GTPCH deficiency), and sepiapterin reductase (SR) deficiency. Generally speaking, early presentation with an array of movement dysfunction is the rule in these disorders, and this umbrella is likely to expand as more disorders are recognized, and more parents join the ranks of the PND Association.

The conference brought together scientists and clinicians to discuss current and potential treatments for PNDs, and to develop practical guidelines for multidisciplinary treatment of these disorders. The conference was attended by more than sixty medical professionals representing six countries, ten medical professionals from the National Institutes of Health, and more than thirty families whose child (or children) are affected with one of the PNDs. As it was with the first Symposium, the opening talks presented an overview of the phenotypes and metabolic features of the PNDs. The future prospects for gene therapy in this group of disorders was also discussed. As always, the exceptionally insightful comments of Prof. Segawa, who in 1971 first described autosomal-dominant GTP cyclohydrolase deficiency, provided thoughtful discussion for all in attendance. A goal of the second Symposium was to present a "multifaceted" approach to treatment. The corresponding sessions highlighted traditional physiatry, sensory integration intervention and the potential for deep brain stimulation for amelioration of symptoms. These approaches generated lively discussion, and it is probable that these approaches will be discussed in future Symposia.

The second Symposium afforded the opportunity for research updates by investigators funded through the PND Association, as well as other investigators who had important new data to present. An impressive update on neuroimaging in AADC deficiency was developed into one of the written contributions to the Symposium proceedings. One of the more rewarding

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aspects of the Symposium, as in the past, was that all of the professionals remained for the final meeting session in which families had the opportunity to sit and discuss their personal situations. The families were extremely grateful for this rare opportunity and the consensus was that professionals and families gained much from these valuable interactions. In total, there were eight reviews presented for publication, representing a permanent record of the second Annual Symposium.

The role of parents and patients in propelling the research enterprise forward is expanding significantly each year. In the past, the relationship between the physician and the patient/family was formal and distant. These relationships have changed more recently, and the benefits of a strong partnership are now abundantly clear. The origins of the Pediatric Neurotransmitter Disease Association are emblematic of this partnership. We would like to acknowledge the indefatigable efforts of four parents from the PND Association whose ongoing support has been instrumental. These champions are Brad and Carolyn Hoffman, and Nancy and John Speller. The proceedings of this Symposium form a testimony to their extraordinary commitment, both to their children and to the PND Association.

The first Symposium was developed simply because so few individuals (and scientists) knew of these rare PNDs. The second Symposium, six years later, was brought to fruition because knowledge of the disorders had expanded, but approaches to treatment were not consistent or sufficiently developed. This time interval between these two Symposia is representative of the slow progress in medicine and the basis for the impatience of parents and patients as they wait for a clearer understanding of the disease that affects them or their children. It is our hope that a future third Symposium, in the not too distant future, will bring effective treatments or, miraculously, a cure for one or more of these disorders. The sponsors of the two Symposia, and the Guest Editors, wish to express their sincere thanks to the *Journal of Inherited Metabolic Disease* for their willingness to report the outcomes of our meeting.