Child Neurology: Autism as a model

Considerations for advanced training in behavioral child neurology

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ABSTRACT

In this article, we advocate for advanced training for child neurologists in behavior and development in order to facilitate the investigation of childhood behavioral and neurodevelopmental disabilities, with autism serving as a model disorder. We explore the current training options and then propose alternative subspecialty training options that focus on behavior and development, with appreciation that most developmental disabilities are not static encephalopathies but, rather, dynamic processes representing the influence of genetics and environment on neural circuitry.

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Both the incidence and recognition of developmental disabilities are growing, with an estimated 9 million children and adolescents affected with autism spectrum disorders, global developmental delay, or mental retardation.¹ Due in large part to increased awareness and earlier diagnosis, the incidence of autism has risen substantially in the past decade, with current estimates being 1 in 150 children.² As evidenced by the increase in NIH funding and media coverage, autism has become a major public health issue. Extensive efforts are being made to understand its neurobiology in order to create informed therapies.

As a new generation of child neurologists is trained, the autism field is turning heavily toward advanced neuroimaging and genetics. Such technological advances, while invaluable, must be matched with sophisticated phenotyping grounded in behavior and development. We submit that well-trained child neurologists with a solid foundation in behavioral neurology would be well-equipped to develop this type of rigorous phenotyping. Coupling clinical observation with an understanding of functional neuroanatomy, behavior, and normal brain development, child neurologists could define phenotypes informed by functional neural networks, which, in turn, would enable us to design effective therapies for these children.

In this article, we advocate for advanced training for child neurologists in behavioral child neurology in order to facilitate the investigation of childhood behavioral and developmental disabilities, with autism serving as a model disorder.

BACKGROUND: THE NEUROLOGY OF AUTISM The concept of approaching autism from a neurologic perspective is by no means a novel one. In the 1970s, Damasio and Maurer³ took a traditional, lesion-based approach in their analysis of the behavioral and motor disturbances in children with autism. Based on their observations, they wrote a seminal paper in *Archives of Neurology* entitled "A neurological model for childhood autism."³

An elegant example of their approach lies in their account of the motor disturbances, characterized as "dystonia, bradykinesia and hyperkinesias and involuntary movements," many of which, they concluded, seemed rooted in the striatum and its connections to the frontal lobe. They provided critical observation that the areas affected "constitute the entire area of termination of the dopaminergic neurons arising in the mesencephalon."³

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Since the localization efforts of Maurer and Damasio, several clinicians and scientists have investigated autism from a child neurology perspective. Through their work, and that of their contemporaries, we have learned much more about the neurologic comorbidities of autism, such as epilepsy, and have better defined autistic regression as distinguished from Landau Kleffner syndrome. We also have learned that several neurologic disorders are associated with a higher incidence of autism, such as tuberous sclerosis complex and fragile X,⁴ and that behavioral syndromes, such as obsessive compulsive disorder and attention deficit hyperactivity disorder, occur at higher rates in children with autism.⁵ Since 2000, 30 original articles about autism have been published in *Neurology*[®], and over 200 in the *Journal of Child Neurology and Pediatric Neurology*.

LOOKING TO THE FUTURE Clearly, the precedent has been set for child neurologists to take initiative in the investigation and care of autism spectrum disorders, but our training must expand upon these efforts. We propose 3 areas of investigation in which child neurologists can make contributions to this field: 1) creation of neurologically based endophenotypes; 2) analysis of early behavioral markers that precede a formal diagnosis; and 3) understanding of the developmental trajectory of autism, focusing on late life sequelae.

Several examples of neurologic endophenotypes can be given. First, neuropathologic and imaging studies have indicated abnormalities in cerebellar volume and structure in children with autism.⁶ In this subgroup, one could ask whether there is evidence of hypotonia, ataxia, or speech impairments attributable to cerebellar circuits. Second, per Maurer and Damasio, one could ask whether children with more prominent movement disorders show evidence of abnormalities in dorsal striatal circuitry as visualized in functional or structural imaging. Perhaps dopamine receptor or transporter genes show mutations that could further refine this clinical profile. Finally, one could ask whether children with autism with specific EEG abnormalities share a common behavioral phenotype, thereby providing insight into common aberrant neural circuitry.

The 2 other areas of investigation (early markers and late sequelae) speak to the importance of understanding autism from a developmental perspective. Autism is not a static encephalopathy, but, rather, a dynamic process that begins in early brain development and continues throughout childhood and possibly later. Observational studies reveal that children with autism can exhibit deficits in social interaction, language, and motor skills in the first year of life, well before a formal diagnosis is made.⁷ Adults with au-

tism often cannot live or work independently, and have a high rate of comorbid psychiatric and behavioral disorders. Furthermore, early childhood language ability is an important predictor of independent functioning in adults with autism.⁸ This dynamic process is substantiated by studies showing dysregulation of brain growth based on head circumference measurements in children with autism.⁹

Child neurologists who can understand behavior in the context of development can investigate autism across a lifetime, from early infancy into adulthood. Through this process, one could ask questions about early precursors of autism, predictors of specific phenotypes, and association of early behavioral and neural markers with late life prognosis. This approach could facilitate the creation of interventions that might modify the developmental progression of autism.

We are using autism as a model disorder, but these suggestions could easily be applied to other, later-onset neurobehavioral disorders, such as schizophrenia, in which early childhood behavioral or cognitive markers may exist.

CURRENT TRAINING OPTIONS Currently, child neurology training consists of 2 years of general pediatrics, 1 year of adult neurology, and 2 years of child neurology. There is a required elective in child and adolescent psychiatry, and most programs offer electives in neuropathology and neuroimaging. While residents gain some exposure to behavioral disorders through outpatient clinics and didactics, there is no formal training in behavioral disorders, psychopathology, or development. In order to gain formal advanced training in behavioral disorders, child neurologists must pursue a fellowship in adult behavioral neurology, for which there is an American Board of Psychiatry and Neurology (ABPN) board certification.

There also exists an accredited Neurodevelopmental Disabilities (NDD) residency which focuses on the longitudinal management of patients with neurodevelopmental disorders. This 6-year residency program was created through joint efforts of the American Board of Pediatrics and the ABPN and consists of 2 years of pediatrics, 1 year of adult neurology, 18 months of clinical child neurology and developmental disabilities, and 18 months of "clinical and basic science" including electives in child psychiatry, neurosurgery, neurorehabilitation, and dedicated research time. Residents are eligible for certification in Pediatrics, Neurology with Special Competence in Child Neurology, and Neurodevelopmental Disabilities.¹⁰ This training is limited to 7 established NDD programs across the country (www.acgme.org).

PROPOSAL FOR TRAINING We envision 2 viable options for training in behavior and development. The ABPN currently provides no mechanism for child neurologists to integrate into the NDD training. Therefore, one advanced training option would be to allow those who have completed their child neurology residency to spend 12–18 months in an NDD program completing its training requirements, with focus on research and clinical electives relevant to developmental and behavioral disorders. This option would be particularly relevant for those who decide to pursue this area of interest after beginning child neurology training. These neurologists could sit for both Neurology and NDD boards.

An alternative option would be to create an accredited fellowship in behavioral child neurology, ultimately with board certification. A 1-year fellowship in behavioral child neurology would include a clinical, didactic, and research component, promoting the understanding of behavior from a developmental perspective. Clinical focus would be placed on children with behavioral and developmental disabilities. As with NDD training, fellows would be given clinical instruction in advanced neuropsychological assessments. Didactics would include lectures on normal child development, psychopathology, childhood behavioral disorders, and psychopharmacology. Advanced instruction in neuroimaging, electrophysiology, or genetics would be provided based on an individual's interests.

Both of these training options would differ from the Developmental Behavioral Pediatric fellowship training, as the latter is a pediatrics subspecialty not focused on neurology or neuroscience. Some of the behavioral issues emphasized in the DBP training, such as encopresis and attachment disorders, are not basic to neurology training and would not be a focus in our proposed training paths.

Finally, we emphasize that all child neurology trainees would benefit from some additional training in neurodevelopmental disorders, particularly as these disorders become more widely recognized and diagnosed. Our proposed training options would provide added benefits to all child neurology residents because of stronger available clinical programs and faculty involvement in development and behavior within child neurology programs.

CONCLUSION It is time for child neurologists to make scientific contributions to this growing field of

neurobehavioral and developmental disorders, with rigorous training opportunities to be able to do so. Armed with knowledge of development, behavior, and clinical neurology, behavioral child neurologists and their NDD counterparts can make innovative, clinically relevant contributions to the assessment and care of children with developmental disabilities and, in the process, regain expertise in the neurology of behavior.

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DISCLOSURE

Dr. Jeste serves as an editorial team member for the *Neurology*[®] Resident and Fellow Section. Dr. Urion has received travel expenses and honoraria for other activities not funded by industry; is an editor of the *Journal of Religion, Disability, and Health*; receives royalties from publication of *Compassion as a Subversive Activity* (Rowman and Littlefield, 2007); and served as an expert witness for a Cleveland Clinic defendant. Dr. Friedman has received travel expenses and speaker honorarium for activities not funded by industry.

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