FOREWORD

The Diagnostic and Statistical Manual of Mental Disorders (DSM; American Psychiatric Association, 2000) specifies that Autism Spectrum Disorder (ASD) is a complex neurodevelopmental disorder that comprises a group of conditions which include the following: Autistic disorder, Asperger disorder, childhood disintegrative disorder, Rett's disorder, and pervasive developmental disorder not otherwise specified. The Center for Disease Control (CDC, 2012) reports that ASD is increasing rapidly with more than one percent of the global population having this disorder. Over the past twenty years, many advances have been made in examining the causes for the disorder; developing accurate identification and diagnostic procedures; validating best practices to instruct children with this disorder; and implementing evidence based social-emotional and behavioral treatment methods (Deisinger, 2012). However, ASD remains a significant worldwide public health issue (Johnson, 2014; WHO, 2013).

Positively, considerable research findings have supported the understanding that ASD is a neurodevelopmental disorder which is characterized by deficits in language and social-emotional functioning and markedly restricted interest and activities (American Psychiatric Association, 2013). These investigations have led to findings that point to the role of genetic factors in the etiology of ASD (Deisinger, 2012). More pointedly, considerable investigations have occurred related to identifying behavioral markers of vulnerability to ASD for the following domains: language, personality, cognitive, and social-emotional. With the advent of new technologies and methods, there has been considerable investigative progress and relevant findings in the past decade regarding knowledge about the association between these behavioral markers and the genetic causes of ASD (Deisinger, 2012). Findings about this association have come from twin and family studies (Hoekstra, Van Beijsterveldt, & Boomsma, 2007), studies about genetic syndromes linked with ASD (Aitken, 2010), and molecular studies of ASD genetics (Aitken, 2010). More importantly, these literature findings point to a constellation of language, cognition, social, and personality characteristics that parallel the defining features of ASD

which are currently being referenced in the literature as Broad Autism Phenotype (Sasson, Lam, Parlier, Daniels, & Piven, 2013).

This volume, The Broad Autism Phenotype, provides readers with in-depth coverage about the broad autism phenotype (BAP). This volume describes in detail the BAP which encompasses biological, cognitive, emotional, behavioral, and interpersonal characteristics resembling those found on the autism spectrum, although more subtle than what is evident among individuals who meet formal criteria for an autism spectrum diagnosis. Initially identified in 1944, the BAP has been receiving increased attention due to the recognition of autism as a spectrum of disorders that vary in symptoms and severity. Researchers believe that studying the BAP may yield helpful information concerning the etiology and early identification of autism.

Chapter 1 of this volume opens with a definition of the BAP, a description of its historical origins, insight into a better understanding of BAP related to its defining elements and boundaries as well as a discussion about a lack of consensus among researchers and professionals regarding what constellation of traits compose the BAP and the variety of measures that are used to investigate it.

Chapter 2 provides a comprehensive examination concerned with the identification and assessment of the BAP. It includes descriptions and critiques of currently used devices and procedures such as interviews, observational approaches, rating scales, checklists, and questionnaires. The chapter author points out while a variety of procedures and devices are available, the detection of the BAP can be challenging due to the lack of uniformity in the way that clinicians and scientists define it, as well as the differing ways that it manifests in males and females. Also, different assessment devices emphasis different aspects of the BAP. As such the chapter's author stresses that the optimal assessment of the BAP should include self reports, information from multiple sources, and direct clinical observations.

Chapter 3 provides a thorough review of genetic aspects of the BAP. The author delineates findings from family and simplex versus multiplex family studies and twin studies. Then, the author discusses endophenotypes which are inherited quantitative phenotypic components of a syndrome (Constantino, 2011). Endophenotypes may be physiological, behavioral, or neuropsychological and are evident in both affected and unaffected individuals (Constantino, 2011). Lastly, the chapter provides a review of molecular genetic studies concerned with the examination of phenotypes due to either gene-gene or gene-environment interactions. The author emphasizes

that the gene-environment aspects associated with BAP is very valuable due to the identification of environmental factors that might be present during prenatal and neonatal development as the identification of these factors would increase the possibility of directing efforts toward the prevention of ASD (Jensen, 2013).

In chapter 4, the author presents information about other biological characteristics of the BAP. The chapter is divided into three sections, namely, differences in brain anatomical structure and function as it relates to the BAP, the relationship between head circumference and the BAP, and the association between the immune system functioning and the BAP. The author points out that recent studies concerned with the above reveal that the BAP involves a variety of biological features that differ from what is found among the general population. These features include: structural differences in both cortical and subcortical regions of the brain which in turn may be linked to atypical activity in the frontal cortex and in areas that are responsible for the processing of social stimuli; and increases in head circumference, levels of whole blood serotonin and testosterone, and rates of obstetric complications. Lastly, the author indicates that while some studies have suggested that autoimmune disorders and epilepsy may be associated with the BAP current research is inconclusive regarding the association between these conditions and the BAP.

Chapter 5 provides an in-depth analysis of the cognitive functioning in the BAP. It focuses on various types of cognitive functioning that have been explored among people with ASD which include the following: perceptual and visual attention, face processing, phonological processing, language ability, intellectual functioning, and executive functioning. In addition, the chapter explores theories more specifically to ASD, namely, central coherence, theories of mind, and social cognition. The author's chapter summary of the available research indicates that individuals with the BAP may engage in atypical processing of sensory information and exhibit unusual patterns of visual orienting and that impaired phonological processing may be an element of the BAP. Further, the intelligence among individuals with BAP is variable and it is unclear whether deficits in executive functioning, central coherence, or theories of mind should be considered characteristics of the BAP. Lastly, studies conducted within the last decade appear to indicate that BAP includes reduced ability for social cognition.

This volume provides autism professionals and researchers with a comprehensive overview of research about the broad autism phenotype from its

earliest recognition until the present time. In addition to an academic readership, the book illuminates information about the broad autism phenotype that may be of interest to family members of individuals on the autism spectrum.

Anthony F. Rotatori
Series Editor

REFERENCES

American Psychiatric Association. (2000). *Diagnostic and statistical manual of mental disorders* (4th ed.). Washington, DC: Author.

American Psychiatric Association. (2013). *Diagnostic and statistical manual of mental disorders* (5th ed.). Arlington, VA: Author.

Aitken, K. J. (2010). *An A-Z of genetics factors in autism: A handbook for parents and carers*. Philadelphia, PA: Jessica Kingsley.

Center for Disease Control and Prevention. (2012). *Prevalence of autism spectrum disorders and developmental disabilities monitoring network, 14 sites, United States, 2008* (Vol. 61, No. 3), Surveillance Summaries.

Constantino, J. N. (2011). The quantitative nature of autistic social impairment. *Pediatric Research*, *69*, 55–62.

Deisinger, J. (2012). Genetic factors associated with autism spectrum disorders. In J. Deisinger, S. Burkhardt, T. Wahlberg, A. F. Rotatori, & F. E. Obiakor (Eds.), *Autism spectrum disorders: Inclusive community for the twenty-first century* (pp. 75–111). Charlotte, NC: Information Age Publishing, Inc.

Hoekstra, R. A., Van Beijsterveldt, T. O., & Boomsma, D. I. (2007). Genetics and environmental covariation between autistic traits and behavioral problems. *Twin Research and Human Genetics*, *10*(6), 853–860.

Jensen, R. A. (2013). The background genetic effect of the genes underlying the broad autism phenotype as a unifying feature in gene x and gene x environment causal mechanisms in autism. *O.A. Autism*, *1*(2), 11–18.

Johnson, E. (2014). Autism spectrum disorders: The worldwide charm and challenge of autism spectrum disorder. In A. F. Rotatori, J. P. Bakken, S. Burkhardt, F. E. Obiakor, & U. Sharma (Eds.), *Special education international perspectives: Biopsychosocial, cultural and disability aspects* (Vol. 27, pp. 117–151). Advances in Special Education. Bingley, UK: Emerald Group Publishing Limited.

Sasson, N. J., Lam, K. S. L., Parlier, M., Daniels, J. L., & Piven, J. (2013). Autism and the broad autism phenotype: Familial and intergenerational transmission. *Journal of Neurodevelopmental Disorders*, *5*(1), 11.

World Health Organization (WHO). (2013). *International Classification of Diseases (ICD) information sheet*. Retrieved from http:www.who.int/classification/icd/factsheet/en/index.html