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Autism and Pervasive Developmental Disorders

Second edition

Edited by
Fred R. Volkmar

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Preface to the second edition

Interest in what is now recognized as the pervasive developmental disorders can be traced to the middle of the nineteenth century with the first descriptions of childhood "psychosis" (Volkmar, 1996). This interest stemmed from an increasing awareness of the importance of the factors of both experience and endowment in child development. Early descriptions of childhood "insanity" were followed by descriptions of childhood schizophrenia (DeSanctis, 1906). The latter term became synonymous with all forms of severe mental disorder in children. The particular genius of Leo Kanner was reflected in his description in 1943 of the syndrome of infantile autism, which he initially believed to be quite different from the forms of childhood "psychosis" then recognized. In the subsequent six decades autism has been the focus of considerable interest from clinicians and researchers alike so that, for example, a large body of work both on research and intervention is now available (Volkmar *et al.*, 2005).

Autism has been the focus of a very substantial body of work. While major advances have been made in both treatment (NRC, 2001) and research (Volkmar *et al.*, 2005) precise pathophysiological models have yet to be specified. In an attempt to specify such models essentially all the theories of psychology and neurobiology have been utilized. While specification of such mechanisms remains an important, if as yet unrealized, goal considerable accomplishments have been made.

A substantial body of research has established the validity of autism as a diagnostic concept, e.g. on the basis of its characteristic clinical features and course. The neurobiological basis of the disorder has been established and better and more effective treatment methods have been developed. Recent attention has focused on genetic mechanisms in autism as well as on the spectrum of conditions that share some similarities with autism and which are now included in the category of pervasive developmental disorder. Some of these conditions, such as Asperger's and Rett's syndromes, were proposed after Kanner's classic description of autism whereas others, notably childhood disintegrative disorder, were proposed many years before Kanner's work.

This book reflects the considerable progress that has been made in recent years in our understanding of autism and related conditions. In this second

edition the contributors have summarized current knowledge in various areas. The contributors represent various disciplines and provide truly international perspectives on current research in autism and related conditions.

The first chapter provides an overview of current approaches to diagnosis and definition of autism and related conditions. Catherine Lord and I review the development of diagnostic concepts and the rationale for the definitions presently employed in both ICD-10 (1993) and DSM-IV (1994). While these definitions have retained an importantly historical continuity with earlier ones they also reflect advances in knowledge and are based on a large body of empirical data (Volkmar *et al.*, 1994). More importantly the international (ICD-10) and American (DSM-IV) depictions are now conceptually convergent.

Advances in diagnosis have improved methods of case detection and are reflected in the results of more recent epidemiological studies. Such research tells us about the prevalence of these disorders and helps in planning for service delivery. Eric Fombonne provides a helpful summary of present knowledge in this area. As he notes, an even larger group of children has impairments in social interaction, and the characterization of their difficulties remains an important area for future research.

Studies of the cognitive and neurocognitive development of individuals with autism have made it clear that it is necessary to study individuals carefully with different levels of cognitive ability using various approaches. Margot Prior and Sally Ozonoff have summarized the literature on this topic with a particular emphasis on those findings most specific to autism. Their chapter is a masterful summary of a large and diverse literature and outlines areas of work that are of particular interest, e.g. in executive functioning.

Children and adolescents with autism and related conditions exhibit major difficulties in the area of communication. In this second edition we are pleased to have Rhea Paul provide a chapter summarizing what is known both about the fundamental difficulties in this area as well as techniques for intervention. Her chapter addresses an important gap in coverage and will be of great interest to all readers.

The importance of genetic factors in the pathogenesis of autism was relatively underappreciated until quite recently. Somewhat paradoxically the field has gone from a situation in which it was believed that genetic factors had little importance for syndrome pathogenesis to the present situation where it appears that such factors have major importance and where specific genetic mechanisms may indeed be identified in the relatively near future. Peter Szatmari and Marshall Jones' chapter reviews this work and notes the major changes in our

understanding of genetic factors in recent years. As he observes, the issue today is not whether autism has a genetic basis but what that basis is.

The relevance of neurobiological factors in the pathogenesis of autism has undergone a similar transformation. At the time of his original description Kanner (1943) minimized the importance of such factors. Now it appears that such factors may be present in 10% or more of cases (Rutter *et al.*, 1994). While the nature of such associations has been debated (Gillberg and Coleman, 1996) these do suggest important areas for research in underlying pathophysiological mechanisms. Fritz Poutska provides a critical summary of this work and notes areas in which knowledge is still lacking.

Given the severity of autism and associated conditions it is probably not surprising that essentially every conceivable intervention has been attempted. Christopher McDougle and colleagues provide a current review of pharmacological treatments. As noted in their chapter recent work suggests important potential benefits of drug treatments in selected cases as one aspect of a total program of intervention.

Over the past 50 years a considerable body of data has established the centrality of intensive, structured, educational intervention as the bedrock of intervention in autism. This work has its origins both in behavioral psychology and special education. As Sandra Harris notes progress continues to be made in behavioral and educational interventions that have improved the outcome of autism and related conditions. Patricia Howlin summarizes these changes in her review of outcome studies. The child with autism or a related condition presents major challenges for parents, siblings, and other family members. However, the improvement in outcome is a major accomplishment of the past 50 years of clinical work and reflects the substantial gains in implementation of remedial programs.

In the final chapter Lynn Waterhouse, Deborah Fein and Emily Nicholas address the nature of the fundamental social disturbance which characterizes autism. The centrality of social deficits in the definition of this, and related conditions, has repeatedly been emphasized in depictions of the disorder beginning with Kanner's initial description. Despite this we continue to have a rather limited understanding of the nature of social deficits in syndrome pathogenesis. The authors provide an interesting synthesis of work in this area.

This book provides a summary of what is known as well as what remains poorly understood. While researchers and clinicians have learned much over the past five decades of work on autism we continue to have little understanding of the nature of the autistic social dysfunction nor, for that matter, of its underlying neurobiological basis. There are, however, reasons to be hopeful. Advances

in treatment (both behavioral and pharmacological) have led to improved outcomes. Current work underscores the possibility that at least some form or forms of autism may have a strong genetic component and it is possible that some genetic mechanism may be described in the relatively near future; such a finding will advance research in other areas as well, as we begin to understand pathogenic mechanisms more fully. The chapters in this book are a testament to the advances that have been made as well as to the continuing need for research and treatment of these perplexing conditions.

I am grateful to all the chapter authors for their contributions to this volume. In addition I wish to thank the series editor, Professor Goodyer, as well as Pauline Graham and the staff of Cambridge University Press for their help and support.

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