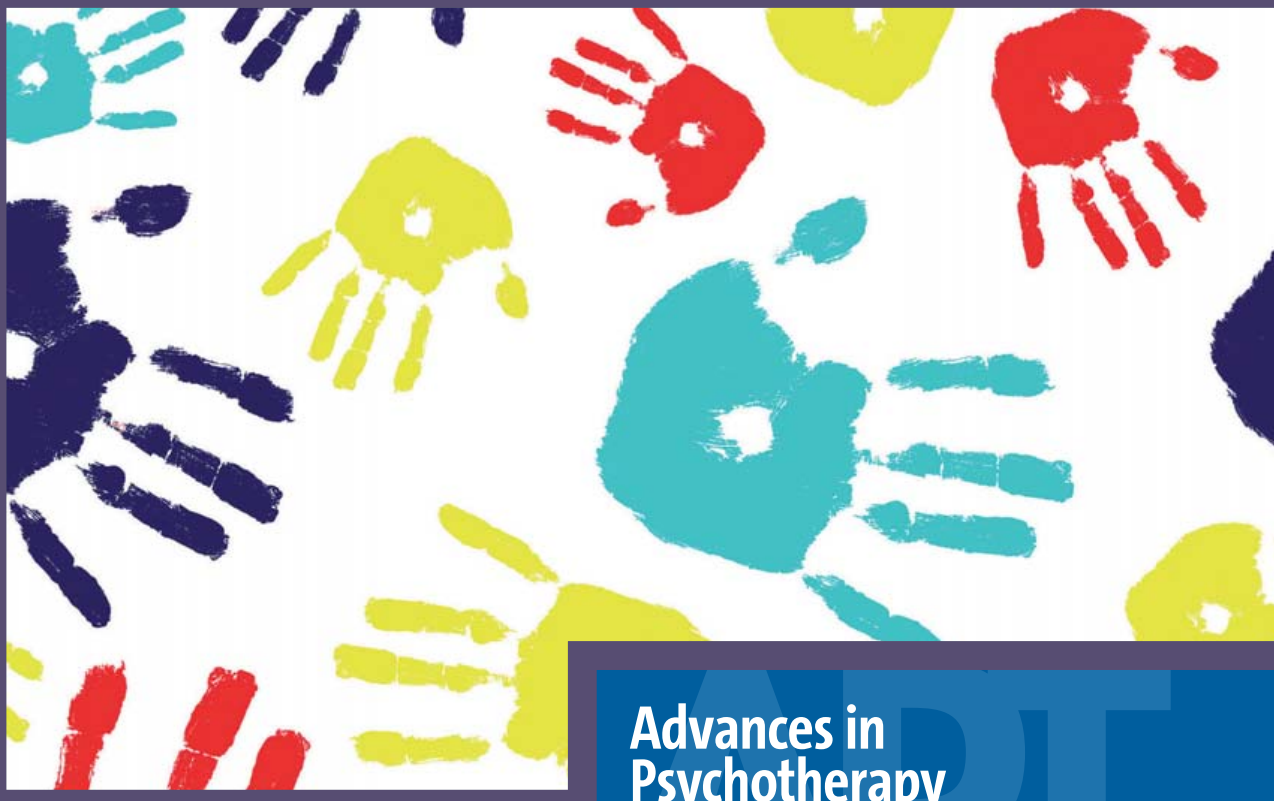
Lisa Joseph · Latha Soorya · Audrey Thurm

Autism Spectrum Disorder



Advances in Psychotherapy

Evidence-Based Practice

HOGREFE

Autism Spectrum Disorder

About the Authors

Dr. Lisa Joseph is a child clinical psychologist. She completed postdoctoral fellowships at Brown University and at the Pediatrics and Developmental Neuroscience Branch of the National Institute of Mental Health Intramural Resarch Program in Bethesda, MD, working with children with autism spectrum disorder (ASD) and other developmental disabilities. Her research interests include repetitive behaviors in children with ASD and differential diagnosis in ASD.

Dr. Latha Soorya is a clinical psychologist and assistant professor of psychiatry at the Autism Assessment Research Treatment and Services (AARTS) Center at Rush University Medical Center (RUMC), Chicago, IL. Dr. Soorya is a clinician–researcher with dual expertise in the evaluation of and evidence-based behavioral interventions for individuals with ASD. Her research interests are focused on improving the efficacy and delivery of outpatient treatments for individuals with ASD across the lifespan. Active research studies include treatment studies targeting socialization and neurocognitive deficits in children with ASD.

Dr. Audrey Thurm is a child clinical psychologist and staff scientist in the Pediatrics and Developmental Neuroscience Branch of the National Institute of Mental Health Intramural Research Program in Bethesda, MD. Her research focus is ASD, and she is especially interested in early diagnosis. Active research studies include natural history studies of autism and related genetic disorders, as well as studies of infants at risk for developing ASD.

Advances in Psychotherapy – Evidence-Based Practice

The basic objective of this series is to provide therapists with practical, evidence-based treatment guidance for the most common disorders seen in clinical practice – and to do so in a reader-friendly manner. Each book in the series is both a compact "how-to" reference on a particular disorder for use by professional clinicians in their daily work, as well as an ideal educational resource for students and for practice-oriented continuing education.

The most important feature of the books is that they are practical and easy to use: All are structured similarly and all provide a compact and easy-to-follow guide to all aspects that are relevant in real-life practice. Tables, boxed clinical "pearls," marginal notes, and summary boxes assist orientation, while checklists provide tools for use in daily practice.

Autism Spectrum Disorder

Lisa Joseph
Pediatrics and Developmental Neuroscience Branch, Intramural Research Program, National Institute of Mental Health, Bethesda, Maryland

Latha Soorya
Department of Psychiatry, Rush University Medical Center, Chicago, Illinois

Audrey Thurm
Pediatrics and Developmental Neuroscience Branch, Intramural Research Program, National Institute of Mental Health, Bethesda, Maryland

HOGREFE

Library of Congress Cataloging information for the print version of this book is available via the Library of Congress Marc Database

Library and Archives Canada Cataloguing in Publication

Joseph, Lisa, author
Autism spectrum disorder / Lisa Joseph, Alliant International University, Hong Kong, China, Latha Soorya, Autism Assessment Research Treatment and Services (AARTS) Center at Rush University Medical Center (RUMC), Chicago, Illinois, Audrey Thurm, Pediatrics and Developmental Neuroscience (PDN), National Institute of Mental Health (NIMH), Bethesda, Maryland.

(Advances in psychotherapy--evidence based practice series ; volume 29)
Includes bibliographical references.
Issued in print and electronic formats.
ISBN 978-0-88937-404-1 (pbk.).--ISBN 978-1-61676-404-3 (pdf).--
ISBN 978-1-61334-404-0 (html)

1. Autism spectrum disorders--Diagnosis. 2. Autism spectrum disorders--Treatment. I. Soorya, Latha, author II. Thurm, Audrey, author III. Title.
IV. Series: Advances in psychotherapy--evidence-based practice ; v. 29

RC553.A88J675 2014 616.85'882 C2014-904947-1
C2014-904948-X

http://www.hogrefe.com

PUBLISHING OFFICES
USA: Hogrefe Publishing Corporation, 38 Chauncy Street, Suite 1002, Boston, MA 02111
Phone (866) 823-4726, Fax (617) 354-6875; E-mail customerservice@hogrefe-publishing.com
EUROPE: Hogrefe Publishing GmbH, Merkelstr. 3, 37085 Göttingen, Germany
Phone +49 551 99950-0, Fax +49 551 99950-425; E-mail publishing@hogrefe.com

SALES & DISTRIBUTION
USA: Hogrefe Publishing, Customer Services Department, 30 Amberwood Parkway, Ashland, OH 44805
Phone (800) 228-3749, Fax (419) 281-6883; E-mail customerservice@hogrefe.com
UK: Hogrefe Publishing c/o Marston Book Services Ltd, 160 Eastern Ave., Milton Park, Abingdon, OX14 4SB, UK
Phone +44 1235 465577, Fax +44 1235 465556; E-mail direct.orders@marston.co.uk
EUROPE: Hogrefe Publishing, Merkelstr. 3, 37085 Göttingen, Germany
Phone +49 551 99950-0, Fax +49 551 99950-425; E-mail publishing@hogrefe.com

OTHER OFFICES
CANADA: Hogrefe Publishing, 660 Eglinton Ave. East, Suite 119-514, Toronto, Ontario, M4G 2K2
SWITZERLAND: Hogrefe Publishing, Länggass-Strasse 76, CH-3000 Bern 9

Format: PDF

ISBN 978-0-88937-404-1 (print), ISBN 978-1-61676-404-3 (pdf), ISBN 978-1-61334-404-0 (epub)
http://doi.org/10.1027/00404-000

Table of Contents

1

Description

This book focuses on autism spectrum disorder (ASD), which is considered a lifelong neurodevelopmental disorder, qualitatively different from other behavioral disorders that are the foci in this series. As a neurodevelopmental disorder, ASD arises very early in life, often has associated biological and medical conditions, and is not easily treated. In fact, although clearly some individuals improve over time and even "remit," it is considered controversial even today to discuss a "cure" for the disorder.

While ASD comprises persisting and impairing social communication deficits and the presence of restricted and repetitive behaviors and interests, it is much more than that, given the inherent and necessary, evolution-driven social tendencies and proclivities of human nature. Lacking basic skills and motivations regarding socialization leads to the inability to engage in and benefit from many of the activities of life that require such skills: learning, conversing, and engaging in meaningful and mutually beneficial relationships among them.

The following work, beginning with an introduction to old and new classifications of the disorder, explores the parameters of ASD as a heterogeneous condition with associated conditions. The book goes into depth about the complexities of making the diagnosis and differentiating ASD from other related disorders, and describes the state of the science and practice with respect to treating associated features and ultimately the core symptoms of the disorder.

1.1 Terminology

The terminology for ASD relates to its associated conditions and the diagnostic classification of the disorder developed through the *Diagnostic and Statistical Manual for Mental Disorders* (DSM) and *International Classification of Diseases* (ICD) classification systems. A few key abbreviations that are used throughout the book are described in the box below.

Key Abbreviations

AAC	Augmentative and alternative communication
ABA	Applied behavior analysis
ASD	Autism spectrum disorder
DTT	Discrete trial training
EIBI	Early intensive behavioral intervention
EST	Empirically supported treatments
FBA	Functional behavioral assessment
PDD-NOS	Pervasive developmental disorder, not otherwise specified
PRT	Pivotal response treatment
RRB	Restricted, repetitive patterns of behavior, interests, or activities
SCD	Social communication disorder

The terminology for ASD has changed several times since the condition was first introduced as a mental disorder in the 1980s, and most recently it has changed with the publication of the *Diagnostic and Statistical Manual for Mental Disorders,* 5th edition (DSM-5; American Psychiatric Association [APA], 2013). The diagnostic classification has changed from autistic disorder (or autism) to autism spectrum disorder (ASD), and ASD is defined with criteria very similar to the previous criteria for autistic disorder (or autism), pervasive developmental disorder, not otherwise specified (PDD-NOS), and Asperger's disorder, under DSM-IV (APA, 2000). The main changes between the DSM-IV and DSM-5 conceptualization and criteria are the following:

1. The broad diagnostic category is termed *autism spectrum disorder* (rather than the previously termed *pervasive developmental disorder*).
2. ASD is one diagnosis, with specific distinctions for autism vs. PDD-NOS vs. Asperger's disorder (as well as Rett syndrome and childhood disintegrative disorder) removed from the DSM, although these terms are still used clinically and are defined in other classification systems.
3. Within ASD, one domain for social communication deficits is now described (merging the separate criteria for reciprocal social interaction deficits and communication deficits).

In addition, key terms in ASD relate to concepts of the restricted, repetitive behavior domain. In this area, symptoms are now described to include fixated patterns of interests (also described as circumscribed interests or preoccupations) and stereotyped movements (which may include repetitive or idiosyncratic movements). The DSM-5 also includes criteria for hypersensory and hyposensory sensitivities, previously not part of the diagnostic criteria.

Level of support indicators: Requiring support Requiring substantial support Requiring very substantial support

The description of relevant terminology relating to ASD also includes wording for how DSM-5 diagnoses are described more generally. Instead of using a multiaxial system, the DSM-5 uses specifiers, which are descriptors used in the diagnosis to more comprehensively describe an individual. The specifiers for ASD include indication of cognitive functioning (with or without intellectual impairment); language level (with or without language impairment); associations with known medical, genetic, or environmental factors; and classifications that may be used to describe severity for the two criteria domains, categorized by three levels of support. These levels of support are described as requiring support, requiring substantial support, and requiring very substantial support, and these may be used to track change

over time within an individual. Separate specifiers are used to describe severity for each domain (social communication deficits and restrictive, repetitive behavior). As such, while some of the specifiers (e.g., association with a medical illness) may be static for individuals, other specifiers may be modified for individuals as they change developmental stage or potentially respond to treatment.

While DSM-5 included the aforementioned changes, the current ICD-10 classifications remain similar to the DSM-IV conceptualizations, as shown in Table 1 (see http://apps.who.int/classifications/icd10/browse/2010). In addition to classifications for autism (childhood autism), other childhood disintegrative disorder, Rett syndrome, and Asperger's syndrome, the ICD-10 system includes an atypical autism classification, for individuals with onset after the age of 3 years. It also includes subthreshold criteria such that a diagnosis of autism may be ruled out. While the ICD-11 revision is not due to be published until 2015, it remains to be seen how and whether these two classifications systems will realign with respect to ASD.

Table 1
DSM-5 and ICD-10 Diagnostic Criteria for ASD

DSM-5	ICD-10
Autism spectrum disorder (299.00) A. Persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following, currently or by history: 1. Deficits in social-emotional reciprocity 2. Deficits in nonverbal communication behaviors used for social interaction 3. Deficits in developing, maintaining, and understanding relationships Specify current severity: severity is based on social communication impairments and restricted, repetitive patterns of behavior B. Restricted, repetitive patterns of behavior, interests, or activities, as manifested by at least two of the following, currently or by history: 1. Stereotyped or repetitive motor movements, use of objects, or speech 2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior	**Pervasive developmental disorders** **Childhood autism (F84.0)** A. Presence of abnormal or impaired development before the age of 3 years, in at least one out of the following areas: 1. Receptive or expressive language as used in social communication 2. The development of selective social attachments or of reciprocal social interaction 3. Functional or symbolic play B. Qualitative abnormalities in reciprocal social interaction, manifest in at least one of the following areas: 1. Failure adequately to use eye-to-eye gaze, facial expression, body posture, and gesture to regulate social interaction 2. Failure to develop (in a manner appropriate to mental age, and despite ample opportunities) peer relationships that involve a mutual sharing of interests, activities and emotions 3. A lack of socioemotional reciprocity as shown by an impaired or deviant response to other

Table 1 (continued)

DSM-5	ICD-10
3. Highly restricted, fixated interests that are abnormal in intensity or focus 4. Hyper- or hypo-activity to sensory input or unusual interest in sensory aspects of the environment Specify current severity: severity is based on social communication impairments and restricted, repetitive patterns of behavior C. Symptoms must be present in the early developmental period (but may not become fully manifest until social demands exceed limited capacities, or may be masked by learned strategies in later life) D. Symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning E. These disturbances are not better explained by intellectual disability (intellectual developmental disorder) or global developmental delay. Intellectual disability and autism spectrum disorder frequently co-occur; to make comorbid diagnoses of autism spectrum disorder and intellectual disability, social communication should be below that expected for general developmental level	people's emotions; or lack of modulation of behavior according to social context, or a weak integration of social, emotional, and communicative behaviors C. Qualitative abnormalities in communication, manifest in at least two of the following areas: 1. A delay in, or total lack of, development of spoken language that is not accompanied by an attempt to compensate through the use of gesture or mime as alternative modes of communication (often preceded by a lack of communicative babbling) 2. Relative failure to initiate or sustain conversational interchange (at whatever level of language skills are present) in which there is reciprocal to and from responsiveness to the communications of the other person 3. Stereotyped and repetitive use of language or idiosyncratic use of words or phrases 4. Abnormalities in pitch, stress, rate, rhythm, and intonation of speech D. Restricted, repetitive, and stereotyped patterns of behavior, interests, and activities, manifest in at least two of the following areas: 1. An encompassing preoccupation with one or more stereotyped and restricted patterns of interest that are abnormal in content or focus; or one or more interests that are abnormal in their intensity and circumscribed nature although not abnormal in their content or focus 2. Apparently compulsive adherence to specific, nonfunctional routines or rituals

Table 1 (continued)

DSM-5	ICD-10
	3. Stereotyped and repetitive motor mannerisms that involve either hand or finger flapping or twisting, or complex whole body movements 4. Preoccupations with part-objects or nonfunctional elements of play materials (such as their odor, the feel of their surface, or the noise or vibration that they generate) 5. Distress over changes in small, nonfunctional details of the environment E. The clinical picture is not attributable to the other varieties of pervasive developmental disorder; specific developmental disorder of receptive language (F80.2) with secondary socioemotional problems; reactive attachment disorder (F94.1) or disinhibited attachment disorder (F94.2); mental retardation (F70-F72) with some associated emotional or behavioral disorder; schizophrenia (F20) of unusually early onset; and Rett's syndrome (F84.2) **Atypical autism (F84.1)** A. Presence of abnormal or impaired development at or after age 3 years (criteria as for autism except for age of manifestation) B. Qualitative abnormalities in reciprocal social interaction or in communication, or restricted, repetitive, and stereotyped patterns of behavior, interests, and activities (criteria as for autism except that it is not necessary to meet the criteria in terms of number of areas of abnormality) C. The disorder does not meet the diagnostic criteria for autism (F84.0)

Table 1 (continued)

DSM-5	ICD-10
	Rett's syndrome (F84.2) A. Apparently normal prenatal and perinatal period and apparently normal psychomotor development through the first 6 months and normal head circumference at birth B. Deceleration of head growth between 5 months and 4 years and loss of acquired purposeful hand skills between 6 and 30 months of age that is associated with concurrent communication dysfunction and impaired social interactions and appearance of poorly coordinated/unstable gait and/or trunk movements C. Development of severely impaired expressive and receptive language, together with severe psychomotor retardation D. Stereotyped midline hand movements (such as hand wringing or washing) with an onset at or after the time that purposeful hand movements are lost **Other childhood disintegrative disorder (F84.3)** A. An apparently normal development up to the age of at least 2 years. The presence of normal age-appropriate skills in communication, social relationships, play, and adaptive behavior at age 2 years or later is required for diagnosis B. A definite loss of previously acquired skills at about the time of onset of the disorder. The diagnosis requires a clinically significant loss of skills (and not just a failure to use them in certain situations) in at least two out of the following areas: 1. Expressive or receptive language 2. Play 3. Social skills or adaptive behavior 4. Bowel or bladder control 5. Motor skills C. Qualitatively abnormal social functioning, manifest in at least two of the following areas:

Table 1 (continued)

DSM-5	ICD-10
	1. Qualitative abnormalities in reciprocal social interaction (of the type defined for autism) 2. Qualitative abnormalities in communication (of the type defined for autism) 3. Restricted, repetitive, and stereotyped patterns of behavior, interests, and activities including motor stereotypies and mannerisms 4. A general loss of interest in objects and in the environment D. The disorder is not attributable to the other varieties of pervasive developmental disorder; acquired aphasia with epilepsy (F80.6); elective mutism (F94.0); schizophrenia (F20-F29); Rett's syndrome (F84.2) **Asperger's syndrome (F84.5)** A. A lack of any clinically significant general delay in spoken or receptive language or cognitive development. Diagnosis requires that single words should have developed by 2 years of age or earlier and that communicative phrases be used by 3 years of age or earlier. Self-help skills, adaptive behavior, and curiosity about the environment during the first 3 years should be at a level consistent with normal intellectual development. However, motor milestones may be somewhat delayed, and motor clumsiness is usual (although not a necessary diagnostic feature). Isolated special skills, often related to abnormal preoccupations, are common, but are not required for diagnosis B. Qualitative abnormalities in reciprocal social interaction (criteria as for autism) C. An unusually intense circumscribed interest or restricted, repetitive, and stereotyped patterns of behavior, interests, and activities (criteria as for autism; however, it would be less usual for these to include

Table 1 (continued)

DSM-5	ICD-10
	either motor mannerisms or preoccupations with part-objects or nonfunctional elements of play materials) D. The disorder is not attributable to the other varieties of pervasive developmental disorder; schizotypal disorder (F21); simple schizophrenia (F20.6); reactive and disinhibited attachment disorder of childhood (F94.1 and F94.2); obsessional personality disorder (F60.5); obsessive-compulsive disorder (F42) **Pervasive developmental disorder, unspecified** This is a residual diagnostic category that should be used for disorders which fit the general description for pervasive developmental disorders but in which a lack of adequate information, or contradictory findings, means that the criteria for any of the other F84 codes cannot be met

Note. ASD = autism spectrum disorder.

1.2 Definition

Deficits in social communication, including reciprocal social interaction, and the presence of restricted, repetitive patterns of behavior, interest, or activities (RRBs) are the defining symptoms of ASD. The onset of these symptoms must be present in early development, the symptoms must cause impairment in several areas of functioning, and the symptoms must not be better explained by intellectual disability or global developmental delay (APA, 2013).

Social communication deficits are among the earliest indicators of ASD

Social communication deficits are among the first concerns reported by parents (Lord, 1995) and have long been considered to be at the core of ASD. Studies have indicated that reduced eye contact, reduced/atypical affect (Clifford & Dissanayake, 2008), poor response to name, and reduced use of communicative gestures are among the earliest indicators of ASD (Osterling, Dawson, & Munson, 2002). Other impairments in this domain include deficits in social-emotional reciprocity (e.g., reciprocal conversation, sharing of inter-

ests) and impairments in nonverbal communication (e.g., eye gaze, gestural communication, facial expressions of emotion). Deficits in the maintenance of social relationships, difficulties adjusting behavior to different contexts, lack of imaginative play skills, and impairment in, or lack of, peer relationships are also defined as social communication deficits.

RRBs are also core symptoms of the disorder. These behaviors manifest as stereotyped and repetitive motor movements, such as hand flapping, spinning, repetitive use of objects, lining up toys, repetitive speech, and engaging in delayed echolalia. Insistence on sameness, inflexibility with regard to routines, and ritualized speech and behavior are also considered RRBs. Unusual attachment to objects, and hyposensitivity or hypersensitivity to sensory stimuli are also subsumed under this category of behaviors (APA, 2013). While some of these behaviors are observed in typical development, as well as in other psychiatric and genetic disorders, differences in patterns and frequencies have been observed in individuals with ASD compared with other groups (Bodfish, Symons, Parker, & Lewis, 2000).

Diagnosing ASD: Symptoms must cause current impairment Symptoms cannot be explained by intellectual disability or global delay

To meet diagnostic criteria for ASD, an individual must either currently or by history, meet criteria for all three of the symptoms in the social communication domain, and must meet criteria for at least two of the RRBs, also either by current status or history. In addition, impairments in these two domains must affect current functioning. Symptoms also cannot be better explained by intellectual disability or global developmental delay. It is important that developmental (mental) age and age-appropriate norms are considered when determining the extent of impairment, as some symptoms occur at later stages of development (e.g., in peer relationships), and other behaviors can be observed in typical development (e.g., hand flapping).

ASD can be reliably diagnosed in children as young as 24 months, through to adulthood. Diagnostic differentiation, especially both in the very young and in adult populations, can be difficult for many reasons. In early childhood, other developmental delays can be misdiagnosed as ASD, and in older individuals, the reliance on retrospective information regarding early development can be an impediment. See Early Warning Signs and FIRST WORDS Project in Appendix 1.

While the aforementioned criteria define ASD as one diagnostic entity, the presentation can differ substantially across individuals due to variability in core and associated symptoms (e.g., age, cognitive functioning, language ability). These facts impact the use of the term *spectrum* in the diagnosis. This heterogeneity has been observed since the disorder was first characterized by Leo Kanner, and can make diagnosis, understanding of trajectory, and targeting of treatment difficult.

The characteristic heterogeneity in the syndrome was not fully captured in the DSM-IV categorizations. Thus, distinct categories were collapsed into one diagnosis in DSM-5. Further, the PDD-NOS criteria in DSM-IV were vague to the extent that it allowed for a relatively heterogeneous group of children to be represented by this diagnosis. Specifically, DSM-IV diagnostic criteria for PDD-NOS were not precise and allowed for subthreshold symptoms in any one or more of the three core symptom domains (i.e., reciprocal social interaction, and/or communication deficits, and/or presence of RRB) and did not require communication deficits and/or RRB to meet criteria.

If significant RRBs (even by history) are present, a diagnosis of SCD is excluded

It is not yet known if and how the DSM-5 diagnostic criteria will affect prevalence rates of ASD, and how individual diagnoses will be made in practice. Field trial data indicated that many individuals previously diagnosed with autism will meet the criteria, as well as those diagnosed with Asperger's disorder and PDD-NOS (Regier et al., 2013). However, the addition of social communication disorder (SCD) in the DSM-5 may also contribute to changes in diagnostic approach. SCD is defined by difficulties with pragmatic aspects of communication, and includes impairments in the social use of language. However, if significant RRBs (even by history) are present such that a child meets full criteria for ASD, a diagnosis of SCD is excluded. Thus, some individuals diagnosed with PDD-NOS in DSM-IV (who did not have significant symptoms of repetitive behavior and restricted interests) may fall under the new SCD category, while others may fall under the ASD category.

1.3 Epidemiology

Prevalence rates of ASD are reported to be increasing, with several studies indicating alarming rate surges. In this section we will provide a brief overview of the epidemiology of ASD, and discuss some of the challenges that impede epidemiological research in ASD.

1.3.1 Prevalence Rates of ASD

The prevalence rate for ASD in the US is slightly more than 1% and has been increasing

In epidemiological studies across the globe, rates of ASD have ranged broadly. According to the US Centers for Disease Control and Prevention, the current prevalence rate of ASD in the US population was most recently reported to be approximately 11.3 in 1,000 children, or 1.13 % (MMWR, 2012). A study in South Korea reported the highest rates thus far, with prevalence reported to be 2.64 % overall, including 3.74 % in males and 1.47 % in females (Kim et al., 2011). Rates in other parts of Asia have varied from as low as approximately 1 % in China (Sun et al., 2013), to approximately 1.81 % in Japan (Kawamura, Takahashi, & Ishii, 2008). Across Europe, overall rates are reported to be approximately 1 % of the population. While incidence (number of new cases) of the disorder is not well measured and has not necessarily increased, the prevalence rate (total number of cases) has appeared to increase over the past few decades.

Factors Affecting Prevalence Rates

Several factors have influenced the change in prevalence rate, including case ascertainment, case identification, and diagnostic substitution. These issues are important to discuss as they demonstrate how changes in diagnostic criteria, improvements in early identification, and methods of identification can influence case ascertainment and thus estimation of prevalence. Studies have generally used two approaches to case ascertainment: (1) a single-stage method involves reviews of service provider databases and national registries; (2) a multistage method uses record reviews as well, but prevalence is determined by use

of a screening tool or diagnostic checklist. For studies that use screening tools, the sensitivity of the screening tools used is of utmost importance. Due to their nature and intended use, screening tools and methods used to confirm diagnoses in epidemiological studies are vulnerable to, and generally are unable to correct for, the presence of both false positives and false negatives in the population.

Case Identification

The diagnostic criteria for ASD have changed over the years, and as it relates to studies of ASD epidemiology, the broadening of the definition of ASD has likely been related to more identification of cases, and a consequent "rise" in prevalence. As the definition of ASD has broadened, awareness of the disorder has increased, and estimates of prevalence have increased. To exemplify this relationship, one review of epidemiology studies reported a positive correlation between prevalence rate and year of study publication (Fombonne, Quirke, & Hagen, 2011).

Diagnostic Substitution

The potential that diagnostic substitution – the change from one diagnosis to another diagnosis over time – has occurred plagues epidemiological studies in ASD. In ASD, this may relate to children who were initially diagnosed with other disorders such as anxiety disorders or intellectual disability, but as diagnostic criteria or diagnostic processes have changed, they may have later been diagnosed with ASD. Conversely, for children who were previously diagnosed with ASD, but who changed or improved over time, an ASD diagnosis may no longer be appropriate. As the field moves toward improving diagnostic evaluations for young children, it is possible that increasing percentages of children with ASD will show instability in diagnosis, which will have implications for prevalence rates.

Demographic Trends

ASD is four times more prevalent in males than females

ASD is approximately four times more prevalent in males than females. This trend has been repeatedly observed across epidemiological studies over the years. There is some evidence that females with ASD are disproportionately more likely to be diagnosed with comorbid intellectual disability, compared with males. Additionally, sex differences in overall symptom severity have not been consistently found (Zwaigenbaum et al., 2012); however, certain types of symptoms and the timing of the identification of symptoms differ by sex (Begeer et al., 2013).

Several hypotheses have been offered for the sex differences observed in the disorder. Recent studies have postulated that females may have sex-linked protective factors that result in decreased prevalence compared with males; further, affected females may have more risk factors than males (Robinson, Lichtenstein, Anckarsater, Happe, & Ronald, 2013).

Racial, Ethnic, and Socioeconomic Status Findings

In general, racial, ethnic, and socioeconomic status (SES) differences in overall diagnosis of ASD have not been found. However, despite the push for early identification, in the United States, communities often differ significantly regarding the age of diagnosis and the extent of the services received. In the United States,

children with lower SES, children from racial and ethnic minorities, and children living in rural communities have a later age of diagnosis, and thus receive intervention services later. In the United States, African American and Hispanic children were reported to be diagnosed 1.5 years later on average than White children (Mandell, Listerud, Levy, & Pinto-Martin, 2002). A review of medical records further confirmed this finding, indicating that children who were African American, Hispanic, and Asian, despite meeting criteria for an ASD, had fewer documentations of a previous ASD diagnosis than White children (Mandell et al., 2009). This disparity in timing of diagnosis also extends to the types of follow-up and subspecialty services received; diagnosis and treatment for comorbid symptoms were reported to be significantly lower in minority populations in the United States (Broder-Fingert, Shui, Pulcini, Kurowski, & Perrin, 2013).

Racial, ethnic, and SES differences in overall diagnosis of ASD have not been found

Several reasons for the disparity in timing of diagnosis have been put forth, with accessibility to services being the front-runner. Early diagnosis in the United States has been promoted because of the benefits of early intervention. However, in some communities, intervention services are either unavailable or difficult to access. Race, ethnicity, and SES can impede identification of ASD and the ability to receive services, resulting in case ascertainment bias in epidemiological studies, which can affect estimates of prevalence. Studies using national registries are limited to families who are in the registry; thus, individuals with limited access to services may remain unidentified.

In the United States and globally, the influence of culture may also result in later identification. Different cultures have a different understanding and expectations of both developmental milestones, and mental health disorders (Grinker, Yeargin-Allsopp, & Boyle, 2011). Further, a main impetus for early identification has been the effect of early intervention; in countries without this type of service infrastructure, it is arguable that the need for early identification may not be as well highlighted.

Another factor that researchers will have to explore is how the course of the disorder affects prevalence rates. Early diagnosis and early intervention can lead to improved outcomes for individuals, and for some, significant reductions in symptoms.

1.4 Course and Prognosis

The course and prognosis of ASD are as heterogeneous as its initial presentation. Characterized as a neurodevelopmental disorder, ASD when diagnosed implies impairment throughout the lifespan, and the majority of individuals with ASD maintain this diagnosis as they age (Lord et al., 2006). The symptom presentation has been examined both cross-sectionally and longitudinally; longitudinal studies provide a context for better understanding current functioning, approaches to intervention, and the trajectory of ASD. In this section, what is known about the stability of the disorder over time, and predictors that lead to positive prognosis, will be reviewed. Understanding the course and prognosis for ASD is complicated by the varying presentations of the disorder, and the presence of associated symptoms. See Appendix 1 for a link to the Centers for Disease Control and Prevention website.