NURSING OF AUTISM SPECTRUM DISORDER
EVIDENCE-BASED INTEGRATED CARE ACROSS THE LIFESPAN

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Nursing of Autism Spectrum Disorder
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This book is dedicated to our families.

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Many of us have had a time when we have struggled with an illness and trusted a nurse to guide us through tests, to provide important information, or just to offer an empathetic ear. For routine care, we trust our health care providers to check up on us and tell us whether things are going well or something about our health has gone awry. We make calls, wait for our appointments, fill out checklists, answer questions, put on the gowns, and comply when asked to “stand here,” “take a deep breath,” and “open up and say ‘aah.’” These encounters might be routine or life-changing. Having that care is something we often take for granted when we go to the doctor’s office. Most of us count on being able to report our history and symptoms and trust the people we encounter are there to help us.

Now, what if you were one of the 1% of people who has an autism spectrum disorder (ASD)? How would that change your health care encounter? From early on, your parents might have had a nagging feeling that something was different. They might have questioned whether you could hear and wonder why you did not turn around when they called your name in that sweet baby voice they used to get your attention. At a 15-month well-baby check, your mother might have kept a close eye on the nurse for any sign that something was different. She might have apologized when you cried and arched your back as you were examined. She might have been anxious and relieved at the same time when asked to answer questions about how many words you were using, whether you smiled when others smiled at you, whether you pointed out objects, and whether you pretended to talk on the phone. You might have been frustrated that you could not say you were thirsty, that people kept interrupting the way you looked at the wheels on a toy car, and that the sound of the air conditioner was scary and sharp to your ears.

From early in life, when the social, communication, and behavioral signs of autism emerge, life is different for people with an ASD. In addition to these core aspects of ASDs, people with an ASD are likely to have co-occurring conditions, such as problems with sleeping, eating, pain sensitivity, attention, and anxiety, among others. Getting routine health care can be a challenge for many reasons, such as the need to communicate and interact with other people. People with an ASD may find the unpredictability of office visits to be disconcerting. The unfamiliar people with unclear intentions in the midst of strange sights, smells, and sounds may be overwhelming. Some people with an ASD may react with outbursts that are frightening to those who do not know them, or they may just shut down and not respond. A person with an ASD may not be able to tell you where it hurts or that he or she even has pain in the first place. Even before the appointment, it may take extra steps and preparation to get ready for an office visit. Additional support and patience throughout the visit may be necessary. In these busy times, many health care professionals may not be interested in taking the time necessary to address the special needs that someone with an ASD may have.

Given the current prevalence estimates and improvements in awareness of ASDs, it is very likely that most health care offices will have patients on the spectrum at some time or
other. Ideally, each person has access to a Medical Home service that provides accessible and family-centered care across the lifespan. People with ASDs may have unique developmental challenges, but they also face a range of health issues common to all people. Many of these may be compounded by autism. For too many, basic health care needs have been overshadowed by autism, with potentially treatable health issues explained away as just another part of autism. There is a need to change that standard, so that each person with ASD has access and is involved in what is needed to help him or her live a healthy life.

This volume, edited by Ellen Giarelli and Marcia R. Gardner, is a much-needed resource for nursing professionals. Nurses are a vital part of the health care team, and information and tools are essential for improving the well-being of people with ASDs. This book helps move health care forward by recognizing the range of issues across the lifespan that people with ASDs may face. In addition, the chapters address nursing care across settings, such as health care, educational, and specialized treatment facilities. This volume brings together a wealth of expertise, with information and strategies across the lifespan. The authors not only address issues of early identification but also issues of safety, nutrition, intervention, and care coordination. Although ASDs are typically lifelong conditions, little has been written about issues that affect adults with ASDs, particularly those who are elderly. The authors also address key transitions, milestones, health crises, and end-of-life issues. Each chapter provides practical illustrations and guidance to help prepare nurses for working with people with ASDs.

The editors have been dedicated to improving the information that drives health care for people with developmental disorders. This book provides much-needed information that follows reports from the Centers for Disease Control and Prevention on the prevalence of ASDs. It addresses practical issues in nursing care, such as evaluating a model for community-based pediatric screening for ASDs among toddlers. This resource will help spread important information to improve the lives of people with ASDs. Through these efforts, I hope that more people with ASDs will feel they are part of a trusted team when faced with being asked to “open up and say ‘aah.’”

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The findings and conclusions presented here are those of the author and do not necessarily represent the official position of the Centers for Disease Control and Prevention.
People with autism spectrum disorder (ASD) seek health care in pediatric primary-care offices, health-screening centers, emergency rooms, and outpatient and specialty clinics. Regardless of their work environment, nurses will inevitably encounter a patient with ASD.

At this time, ASD has no cure. There are treatments designed to mitigate the effects of symptoms and core and associated features. The core and associated features of the disorder may interfere with, or prevent, a patient’s receipt of expert, quality, compassionate health care and cure for conditions that require nursing and medical treatments. This book does not discuss standard treatments for ASD; it presents issues surrounding lifelong, comprehensive care for people with ASD. Reading this book will prepare nurses to recognize the unique circumstances of providing health care to any person on the spectrum. The content assists nurses in organizing their care in ways that ensure any patient with ASD they encounter will receive expert, evidence-based care, access to appropriate medical interventions, and an equal opportunity to achieve optimal functioning and the highest possible quality of life. To accomplish this, a nurse must systematically apply, to each clinical encounter, knowledge of the typical behavioral characteristics of individuals with ASD; information on the individual’s unique idiosyncrasies; and the person’s stage of physical, mental, and emotional development. This will result in the delivery of intelligent, authentic, sympathetic, and deliberate care.

AN ORGANIZING PRINCIPLE

One organizing principle of this book is that of a social model of disability. In the broadest sense, a social model of disability is concerned with a clear focus on the environmental, cultural, and economic barriers encountered by people who are viewed as having some form of physical, mental, or intellectual impairment. The barriers encountered include: inaccessible educational systems and working environments; inadequate disability benefits; discriminatory health and social services; inaccessible transport, housing and public buildings, and amenities; and the devaluing of disabled people through negative images of those who are differently abled (Oliver, 2004).

Since its description over 60 years ago, autism has puzzled researchers and clinicians and generated scientific research across a wide range. Autism was under the purview of pediatric psychiatrists and other mental health professionals, who observed similarities with schizophrenia, thought disorders, and mental retardation. Psychogenetic explanations dominated the literature until a broader conceptualization of autism widened the view to include those with extremely high intelligence and unique talents and skills.

Until recently, children were the major recipients of medical and social attention. However, the increased awareness among parents, teachers, doctors, nurses, and the public has contributed to the development of a more comprehensive and integrated approach to care. Such an approach requires inclusion of a range of professionals, including nurses.
Moreover, a lifespan approach is necessary, as families have made it clear to all health care providers that ASD is not a disorder of childhood alone, but rather is a lifelong challenge for those who are affected.

Nurses are the largest group of health care professionals and will certainly provide care to many individuals with ASDs. Our case-based approach to patient care demonstrates the application of health history and physical and mental health assessments to the delivery of integrated care that is authentic, sympathetic, and deliberate. A case-study approach captures typical medical situations and demonstrates how nurses can apply care-planning strategies across communities and service settings.

The main themes are:

- ASDs is a growing public health problem and people with ASDs have special health care needs across the lifespan.
- As the largest group of health care professionals, nurses will interface with people with ASDs and their families in every health care setting.
- Nurses must integrate specialized information on ASDs into their clinical practice, across multiple practice settings.
- Setting-specific case studies illustrate “best practice” nursing care for a variety of problems experienced by people with ASDs.

The book is intended as a textbook and reference. It can serve as source of content for a continuing nursing education program or as the required text for a nursing minor in integrated ASD nursing care. It was conceived as the required textbook for a master’s degree–level minor and a post-master’s certificate program for master’s-prepared nurses, it is therefore an excellent clinical resource for pediatric and mental health nurses, family and adult nurse practitioners, school nurses, college health nurse practitioners, and nurses in nurse-managed health care clinics. It is also suitable for post-baccalaureate education in ASD care.

The book is organized by sections and chapters. Each section begins with an overview of the section’s contents and context and concludes with review questions and additional cases that will help a nurse increase his or her ability to apply knowledge to practice and generate creative solutions to clinical problems. Each chapter comprises a case study exemplar and elaboration of key problems, discussion of solutions, and a “best practice” nursing plan of care. The chapter content provides typical cases and comprehensive nursing care. Evidence-based justifications and examples apply science to nursing care in diverse settings: at the bedside, in primary and tertiary care, and in the community. Because ASD is a developmental disability, this book considers an individual’s developmental needs as part of the organizing framework.

Taking a lifespan perspective, the book uses case studies that illustrate ASD scenarios across the lifespan to highlight nursing’s significant role in the care of people with ASDs. To date, clinical emphasis has been placed on screening and early diagnosis of ASD. Although communication, language, and behavioral deficits may be mitigated through behavioral interventions, ASD is a pervasive disability and a chronic condition, and its core features and problems persist throughout life. Nurses will welcome this innovative and application-focused textbook and reference, which supports effective and appropriate nursing care throughout the lifespan.

TIMING

This book arrives at a most opportune point in time. Mental health and other health care professionals in the United States and globally have turned their attention to describing the
trends in prevalence of ASD (Centers for Disease Control and Prevention, 2009) and its etiological factors.

This book responds to public concerns that the rise in prevalence of ASD is not sufficiently met with treatment options and responds to a call from the Obama administration and the Intra-Agency Autism Coordinating Committee to step up research and treatment for people with ASD. Taking an alternative path, this book addresses these needs by alerting nurses to the complex issues faced by people with ASDs as they struggle with this disability from diagnosis to end of life. Designed for nurses and written by nurses, it is unique among the many textbooks that elaborate on behavioral treatments and posited etiologies.

Arriving on the heels of the new statistics on the trends in prevalence of ASD in the United States (Centers for Disease Control and Prevention, in press), this nursing text affirms the profession’s commitment to evidence-based care.

REFERENCES


Any text of this importance and complexity involves the help of many individuals. This text is not an exception. We gratefully acknowledge the support of the Philadelphia Health Care Trust for providing a generous grant to the University of Pennsylvania to develop ways to prepare nurses to care for the population of children and adults with autism spectrum disorder (ASD). We thank Wiley Thomas and Cathy Greenland for tirelessly advocating for a nursing presence in ASD care. With the collaboration of Jean Ruttenberg, Paula Cullinan, and Joseph Lukach and the staff from the Center for Autism in Philadelphia, we were able to observe firsthand the significant contributions nurses can make to the care of people with ASD and their families. We thank Springer for giving us the opportunity to present this unique, vital, and progressive approach to ASD care. We are indebted to Michelle Savard and Andrea Segal for their work proofreading the contents and their editorial prowess. Finally, we acknowledge our colleagues in medicine, nursing, speech-language pathology, occupational therapy, psychology, and education for helping children and adults on the spectrum find ways to be in their world.
Since its discovery, or naming, over 65 years ago (Kanner, 1943), autism has been a curious and fascinating disorder that has engendered intense concern among parents, teachers, researchers, and communities. As a result of careful research and thoughtful observation, some once-held beliefs have been proven wrong. First, autism is not caused by poor parenting. Similar features in parents of autistic children may be due to genetic factors, as is the increased incidence among siblings of affected children (Rutter, 2001). Second, autism is not part of the childhood schizophrenia group (Kolvin, 1971). It is a neurodevelopmental disorder, not a psychosis. Third, autism is not secondary to a developmental receptive disorder (Cantwell, Baker, Rutter, & Mawhood, 1989).

AUTISM SPECTRUM DISORDER IN FAMILY CONTEXT

From the first recorded observations of a person exhibiting autistic behaviors, it has been seen as a “family matter.” The earliest record of autistic-like behaviors in a patient dates back to a legal case reported in 1747 that sought an annulment of marriage of a man who had “deficits in social relationships including tactlessness and abnormal gaze, echolalia, obsessive and repetitive behaviors” and a preference for sameness (Frith, 2003; Houston & Frith, 2000; Wolff, 2004). Many decades passed before “family dynamics” became the target of health care, and many more decades passed before treatment for behavioral problems took a family-centered and integrated approach. By expanding observations of one person’s problematic behaviors to encompass the effect of these behaviors on members of the extended family and the larger social network, a window opened onto the extensive impact of autism spectrum disorder (ASD) on communities. When communities are involved, nurses are involved. Gray (1998) described becoming a family with autism as an insidious process wherein a family experiences a growing awareness of the problem and gradually adapts to the situation. Because the trajectory of the disorder varies, and there is much diversity across the group of affected persons, families have their own ways of confronting the challenges of ASD, as well as different interpretations based on cultural background. Gray (1998) noted that there is “no clear path of referral . . . no hospital emergency room to treat their child and start them on the road to recovery” (p. 23). In the past, families thought the experts had little to offer. Although this has changed with the help of teachers and therapists, autism remains a family problem. Parents are the principal advocates for their children and are often responsible for securing ongoing and consistent care. Therefore, integrated services for people with ASD must
include the family whenever possible. Nursing care of people with ASD considers the family perspective and integrates this with each clinical encounter.

Accurate diagnosis of ASD is critical to early intervention and improved outcomes for children and their families. Once ASD is diagnosed, evidence-based interventions can ameliorate the symptoms. Until recently, the traditional and consistent focus of treatment was on core features that interfered with functional ability. Now diagnostic best practice is formulated as a three-stage approach: Stage 1: first-level screening; Stage 2: second-level comprehensive developmental and medical evaluation; and Stage 3: specialized evaluation involving administration of autism-specific diagnostic tools (Filipek et al., 2000). Superimposed on these stages is a layer of assessment that considers the impact of core features, as well as characteristics and other symptoms, on other aspects of health care, such as preventive and curative medicine for conditions unrelated to ASD.

The salient points for the early identification of ASD are expanded to apply to every clinical encounter (see Table S1.1).

THE POLITICS OF DISABLEMENT

In medical sociology, the term “disable-ism” refers to the social imposition of avoidable restrictions on the life activities and well-being of people described as “impaired” by those self-described as “normal” or typical (Thomas, 2007). Disable-ism stands with sexism, ageism, and homophobia as a form of social oppression and is found both in person-to-person interactions and within the culture of institutions.

The clinical encounter between health provider and patient is one of the most purposeful of social interactions. It is guided by the expectation of the delivery of and the acceptance of care. It is also an encounter during which disable-ism may become manifest if the care is
guided by unrealistic expectations by the health care provider. In other words, in the clinical encounter with a person with ASD, the nature of the patient’s disability must be understood fully in order to avoid and prevent discrimination. All patients have special needs at one point or another in their lives. A patient-centered approach takes inventory of each person’s abilities. This is essential for the ASD population, as one often hears in the ubiquitous phrase about ASD: “if you met one person with ASD you understand one person’s ASD.”

A patient-centered approach begins by thinking critically about the entire process of case management and, at minimum, communication with the individual with ASD. He or she may not look you in the eye and may seem to glance to the side if peripheral vision is better than frontal vision, but chances are he or she is listening and will respond. Idiosyncrasies can be anticipated and can be included in patient-centered treatment plans.

A study by Heidgerken and colleagues (Heidgerken, Geffken, Modi, & Frakey, 2005) of autism knowledge in health care settings reported that primary-care providers and specialists (e.g., psychiatrists, speech pathologists) had different understandings of the natural history, treatment, and prognosis of autism than autism specialists. This translates to a high potential for inconsistent health care across services, settings, and professional practice. People with special needs may have had negative, even traumatizing, experiences with the health care systems due to differences in provider knowledge and skill at interacting with these individuals. This simple fact should guide nurses to adopt a thoughtful process of caring for people with ASD, beginning at the point of screening and diagnosis and followed throughout their care by anticipating problems. Nurses should endeavor to make every patient’s encounter with the health care system a positive experience that promotes wellness.

Autism was once considered incurable, but that notion has lost strength in the light of increased understanding of the disorder. Every day, individuals with ASD are showing us that they can overcome, compensate for, or manage their most challenging characteristics. The significant increase in public awareness of ASD is largely attributed to the tireless advocacy of parent support groups and national organizations, such as Autism Speaks and the Autism Society of America, which were started as early as the 1960s and are now international. Parents who are frustrated with health care services are driven to seek information and support (Baas, 2006). One important aim of Section I is to equip nurses with knowledge sufficient to help people with ASD benefit from the health care services they need and to which they are entitled.

Section I provides an overview of the public health problem of ASD and introduces the role of nurses and the contributions that the nursing profession can make to improve care for this population. Chapter 1 introduces the pervasive developmental disorder of ASD, with the description of the core features, associated features, and medical comorbidities. The features and characteristics are examined further in the context of specific clinical encounters (e.g., case studies). This chapter also presents the history of ASD, patterns of development, and the evolution of diagnostic criteria. Chapter 2 describes the natural history, etiology, rising prevalence, and risk factors of ASD. Changes in prevalence statistics and possible causes of ASD are outlined. Chapter 2 also describes the rise in public attention toward ASD, which has generated scientific inquiry on the disorder’s prevalence and increased concern for the development of effective treatments and measurable outcomes. Regardless of the chief complaint or purpose for seeking medical care, the clinical management of patients with ASD may ultimately depend on the etiology. Outcomes of effective treatment for any health problem are fused, inextricably, with the behavior features of ASD. Chapter 3 addresses the purpose and value of a comprehensive, integrated approach to the clinical management of ASD, the evolving role of nurses in the care of affected people and their families, and the need for a life-span approach to health maintenance, illness prevention, and integrated treatment protocols.
Best-practice nursing care for this population is a consequence of integrating theories of nursing with theories of ASD in the context of the clinical encounter.

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Autism Spectrum Disorder: Core Features and Medical Comorbidities

Cordelia Robinson Rosenberg

HISTORY

According to the Centers for Disease Control and Prevention, the condition called autism spectrum disorder (ASD) affects approximately 1 in every 88 children and has received considerable attention over the last decade (Centers for Disease Control and Prevention [CDC], 2012). Media of all types provide us almost weekly with some new piece of information about the diagnosis, prognosis, or interventions for this complex disorder. The media attention given to the prevalence of ASD, as well as its social and economic impacts, have been largely influenced by the work of parent- and grandparent-driven advocacy efforts. In addition to media attention, these advocates have influenced the U.S. Congress. The Child Health Act passed in 2000 gave considerable impetus to federal action by authorizing initiatives at the CDC, which included autism surveillance, a multisite case-cohort study (Centers for Autism and Developmental Disabilities Research and Epidemiology—Study to Explore Early Development), and a public awareness campaign around screening (Learn the Signs, Act Early [CDC, 2010]).

In 2006, the U.S. Congress passed the Combating Autism Act, which mandated the reactivation of an Interagency Autism Coordinating Committee (IACC) charged with developing and overseeing a strategic plan. This plan was organized around critical questions framed from the point of view of families: (1) When should I be concerned? (2) How can I understand what is happening? (3) What caused this to happen and can it be prevented? (4) Which treatments and interventions will help? (5) Where can I turn for services? (6) What does the future hold? (Combating Autism Act, 2006).

In December 2010, the Congressional Autism Caucus released a report on the status of the strategic plan. This level of government effort around a specific condition is unprecedented. The number of individuals and families affected by autism, the access to comprehensive care, and the predicted economic impact of a failure to attend to the issues raised by the IACC strategic plan appear to be the primary drivers of this bipartisan government action (Ganz, 2007; Kogan et al., 2008).

In this chapter, we examine the history of diagnosing conditions on the autism spectrum. We will examine the evolution of the criteria used to diagnose autism, the symptoms and behaviors considered to be core features, and the frequency of co-occurring conditions. Before considering these topics, we look at where we are now with respect to the diagnosis of ASD.

DIAGNOSING AUTISM

One of the critical aspects of the work to be done in the field concerns how a diagnosis of autism is made. It is essential to realize that there is no definitive biological test at this time
to substantiate a diagnosis of autism. Autism spectrum disorders are diagnosed clinically based on three core behavioral dimensions: (1) communication delays or deficits, (2) impaired social interaction, and (3) repetitive behaviors and restricted interests (American Psychiatric Association [APA], 2000). These three domains of behavior must be looked at in the context of the individual’s age and stage of development.

Individuals who meet clinical criteria for an ASD diagnosis vary greatly and can include people with extremely high IQs who lack adaptive behavior skills, as well as individuals with severe to profound impairments in intellectual development and no prospects for independence. ASD is highly heritable, and extensive research is being done to identify specific genes related to specific behaviors (Abrahams & Geschwind, 2008; see also Chapter 2, this volume). Some researchers are arguing that the effort to identify the genes responsible for autism would proceed more quickly were the triad of symptoms broken apart and careful characterization of individuals with ASD with respect to each core dimension done (Happe & Ronald, 2008).

Given the clinical nature of the diagnosis, practitioners rely on diagnostic taxonomies or conventions for making a diagnosis. In the case of ASD the systems used are the Diagnostic and Statistical Manual from the American Psychiatric Association and the International Classification of Diseases from the World Health Organization. Given these systems physicians and psychologists are the primary practitioners who are likely to make a diagnosis. Technically any licensed physician or licensed psychologist may make a diagnosis of ASD using criteria in either taxonomy. Many practitioners are reluctant to make such a diagnosis in the absence of having extensive experience with individuals with ASD. However, given the increased services and supports available, especially in the toddler and preschool years, and the long waiting lists and distances to be traveled for expert diagnostic services, some physicians and psychologists are going ahead and giving a diagnosis of autism in order to give families access to services that otherwise might not be available to them.

Currently, there is good congruence between the International Classification of Disease-10 (WHO, 1993) and the DSM-IV Text Revision (DSM-IV-TR; APA, 2000) classification systems. Both systems include three disorders in ASD: autism, Asperger’s disorder, and pervasive development disorder—not otherwise specified (PDD-NOS). Both have comparable lists of criteria for the diagnosis. The development of diagnostic tools, such as the, Autism Diagnostic Observation Schedule (Lord et al., 1989), which combines knowledge of an individual’s history, key informant reports, and direct observation have resulted in adequate sensitivity and specificity for the diagnosis of autism in preschool and school-age children.

Stability of the System of Diagnosis

Diagnoses of autism are generally stable. However, a number of factors are challenging the utility of the current systems. There are difficulties in reliably differentiating between autistic disorder, Asperger’s disorder, and PDD-NOS. In some cases, differentiation depends on the clinician’s access to an individual’s history, which may be problematic, especially for adults. In light of such discrepancies, a study group in neurodevelopmental disorders has been working on a revision for the DSM-IV-TR, which is expected to be fully adopted in 2013 (Lord, 2011). While still being tested, there has already been extensive publicity about the proposed changes to be included in DSM-V. The DSM-V proposes the term “autism spectrum disorder,” with no differentiation to be made among autistic disorder, Asperger’s disorder, and PDD-NOS. Rett syndrome and childhood disintegrative disorder, which were included in DSM-IV, are no longer referred to within ASD. The overarching term is to be ASD, whereas in DSM-III
and DSM-IV it was pervasive development disorder. Another major change in DSM-V for ASD is the fact that being diagnosed as meeting criteria of ASD does not exclude being identified with other diagnoses, such as attention deficit hyperactivity disorder (ADHD).

Historically, the symptoms of ASD were characterized in terms of three domains of behavior: social interaction deficits, communication delays and disorders, and behavior problems characterized by restricted and repetitive behaviors that interfere with development. In the proposed DSM-V, symptoms will be categorized in two domains: social communication and restricted and repetitive behaviors (RRBs). The rationale for this change is that social impairment and communication deficits are only moderately correlated in typical populations, whereas social and communication skills are highly correlated within samples of children or adults with ASD (Lord, 2011).

The DSM-V neurodevelopmental disorders work group has also proposed the use of specifiers and modifiers (Lord, 2011). These specifiers and modifiers address issues of intellectual disability, etiology, age of onset, and severity. With respect to intellectual disability, the recommendation is to indicate whether the person with a diagnosis of ASD also meets criteria for a diagnosis of intellectual disability (ID), which is measured intelligence and adaptive behavior of 70 or below on a valid, norm-referenced, standardized assessment tool. If a person has a genetic syndrome, such as Fragile X or Down syndrome, their diagnosis of ASD would read, for example, as ASD with Fragile X. While there are people who object to some of these changes, overall they reflect the research that has been occurring and the need expressed in the field for better documentation of the behavior of individuals and characterization of populations of people with an ASD (Agency for Healthcare Research and Quality [AHRQ], 2011).

As noted, DSM-V does away with a differentiation among autistic disorder, Asperger’s disorder, and PDD-NOS with the rationale that accurate differentiation is not reliable among diagnosticians. When the proposed change to DSM-V was publicized in 2010, there was media attention given to the protests about this change from people who have a diagnosis of Asperger’s disorder (Adams, 2010). Their concern reflected a desire on the part of people with Asperger’s disorder, many of whom have a very high IQ, to not be associated diagnostically with people with autistic disorder, which they see as associated with low IQ.

Initial Identification of Autism

Autism was first described in 1943 by child psychiatrist Leo Kanner (Kanner, 1943). In his article, Kanner described 11 children he characterized as lacking the typical motivation for social interaction (Volkmar, Klin, & Cohen, 1997). One year after Kanner published his work, Hans Asperger, a medical student in Vienna, published a paper that proposed the concept of autistic psychopathology with respect to a group of boys with whom he was working (Asperger, 1944).

Kanner proposed two categories of symptoms: inadequate social responsiveness and perseverative behavior (Kanner, 1943). Kanner’s descriptions of the children and their parents, particularly the mother’s relationship to the child being described as “cold,” led many to attribute autism to behaviors of the parents. Asperger described his cohort as having difficulties in communication, but felt as though they did not have mental retardation (Asperger, 1944). He based this distinction on the fact that many had areas of average or above average performance on some subtests of the intelligence tests administered to them (Asperger, 1944). Kanner’s and Asperger’s descriptions have a number of common features, but Asperger’s descriptions suggested that high IQ was a feature of the disorder. Neither Kanner nor Asperger seemed aware of the other’s work or their common use of the term autism (Volkmar et al., 1997).
Refining the Description and Understanding of Autism

Kanner’s view that the disorder was congenital and that its expression was affected by the quality of the parent–child relationship was the predominant view of autism until the 1960s, when an alternative perspective to the psychogenic view of autism was offered. Rimland (1964) argued that autism was a biologically based disorder. Rimland’s book, *Infantile Autism*, offered a view of autism as a disorder of information processing. Rimland also developed the first widely used scale focused on specific symptoms of autism (Rimland, 1968).

In 1978, Michael Rutter, a psychiatrist, published a definition of autism that included four essential features: (1) early onset by 2.5 years; (2) impaired and distinctive social development; (3) impaired and distinctive communication; and (4) unusual behaviors consistent in many ways with Kanner’s concept of an “insistence on sameness” (Volkmar et al., 1997). Rutter’s work, as well as that of others, laid the groundwork for the inclusion of specific criteria for the diagnosis of autism included in *DSM-III* (APA, 1980). Autism was not named specifically in the first (APA, 1952) or second (APA, 1968) editions of the *DSM*. The diagnostic option in *DSM-I* and *DSM-II* was childhood schizophrenia (Volkmar et al., 1997).

From the initial identification of autism and Asperger’s disorder in 1943 and 1944 through the 1960s and 1970s, autism was still considered a rare disorder. With the work of Rutter and others and the inclusion of the disorder in *DSM-III* (APA, 1980), autism began to be identified more frequently, but still qualified as a rare disorder. Volkmar and colleagues (1997) pointed out that *DSM-III* is noteworthy, as it developed a taxonomy based upon research findings in which emphasis was placed upon assessing the reliability of descriptions of behavior consistent with a clinical diagnosis. In *DSM-III*, the term pervasive development disorder was introduced. The concept was important, because it avoided any assumptions about etiology.

The approach of looking at multiple disorders under the umbrella term of pervasive development disorder persisted through the next three iterations of the *DSM* (*DSM-III* [APA, 1980]; *DSM-III-R* [APA, 1987]; *DSM-IV* [APA, 1994]; *DSM-IV-TR* [APA, 2000]). In *DSM-IV*, five disorders were included under the term pervasive development disorder: Asperger’s disorder, autistic disorder, PDD-NOS, childhood disintegrative disorder, and Rett disorder.

From 1980 to the present, work has been done within the context of the *DSM* taxonomy to refine our understanding of this complex disorder. Parallel to the work on the *DSM*, there have been refinements to the International Classification of Disease diagnostic taxonomy (WHO, 1993). Research has included comprehensive field trials involving both clinicians with extensive experience and those with less experience with the diagnosis of autism. Sensitivity and specificity of examiner’s classifications of individuals were examined using *DSM-III*, *DSM-III-R*, and ICD-10 criteria (Volkmar et al., 1994). In this comparison, researchers found greater reliability using the ICD-10’s more detailed criteria (Volkmar et al., 1994). Investigators also found that greater experience with autism, rather than professional discipline, was a better predictor of high reliability (Volkmar et al., 1994).

The *DSM-IV* and *DSM-IV-TR* (APA, 1994, 2000) were further refined to establish a definition “that balanced clinical and research needs, was reasonably concise and easy to use, provided reasonable coverage over the range of syndrome expression, and was applicable over the full life span, from early childhood through adulthood” (Volkmar et al., 1997, p. 25). Volkmar, Paul, and Klin (2005) argued that the convergence between systems (*DSM* and ICD) created a synergy between research and clinical practice that precipitated advances in our knowledge of the range in expression of ASD.
One of the features of the *DSM* is that it recommends that consideration be given to five axes when making a diagnosis. “The use of the multiaxial system facilitates comprehensive and systematic evaluation with attention to the various mental disorders and general medical conditions, psychosocial and environmental problems, and lack of functioning that might be overlooked if the focus were on assessing a single presenting problem” (APA, 2000, p. 27). Interestingly, mental retardation is noted on the axis that includes personality disorders. Noting the presence and severity of mental retardation, now referred to as intellectual disability, is a critical aspect of a clinical diagnosis of ASD. An individual may have one or more diagnoses or problems on any of the five axes.

## ELEMENTS OF A COMPREHENSIVE ASSESSMENT

The recommended practice for diagnosing ASD includes completing multidisciplinary evaluation. The purpose of the evaluation is not simply a diagnosis, but rather a comprehensive view of the individual’s strengths and weaknesses and any co-occurring conditions. Both the American Academy of Neurology (Filipek et al., 2000) and the American Academy of Pediatrics (AAP; Johnson, Myers, & the Council on Children with Disabilities, 2007) have developed guidelines for practitioners regarding this process. These guidelines are highly valued and generally viewed as the desired practice, although the AAP cautions that this guidance does not “serve as a standard of medical care” (Johnson et al., 2007, p. 183). The AAP calls for three goals to be addressed in the comprehensive evaluation of a child with suspected ASD. They are: (1) the administration of appropriate developmental or psychometric measures; (2) the presence or absence of a categorical diagnosis of ASD, made with standardized tools demonstrated to be valid and reliable for the purpose; and (3) the presence of other evaluations to determine whether there is an associated etiology for the autism, such as a genetic disorder.

Lord (2011) outlined a number of features critical to a comprehensive assessment, which is applicable for all disorders. Early history, including age of perceived onset and pattern of onset should be documented. Pattern of onset refers to whether there is a perceived or documented loss of words and social skills before 18 months or 30 months versus no clear onset and no loss of words (Lord, 2011). Assessment should also include administration of norm-referenced standardized measures of development for young children or verbal and nonverbal IQ for older individuals. Adaptive functioning should be assessed. Verbal abilities at the time of applying the diagnostic criteria should be documented. Adequacy of initiating and maintaining sleep should be evaluated. Emotional self-regulation ability for age should be evaluated. Any co-occurring developmental, medical, and psychiatric problems should be documented. It is expected that this assessment process can benefit from input from a number of disciplines, including medicine, psychology, speech and language pathology, and occupational therapy, as well as others.

### Assessment Tools

As the research on autism progressed, the need for more refined diagnostic criteria became apparent. The Autism Diagnostic Observation Schedule (ADOS; Lord et al., 1989; Lord, Rutter, DiLavore, & Risi, 1999; Lord et al., 2000) and the Autism Diagnostic Interview-Revised (ADI-R; Lord, Rutter, & LeCouteur, 1994) were developed and became more commonly used for research and also for clinical purposes. Lord and Bishop (2010) point out that the current prevalence of ASD reflects a broader range of functioning referred to as the “broader autism phenotype.” Given this broader range, a one-size-fits-all approach to
assessment is not appropriate. However, Lord and Bishop (2010) go on to point out that there does need to be a standard protocol with steps to take depending upon specific results.

The ADOS has four modules that allow for an assessment of people with different levels of functioning. The ADI-R provides important information regarding the person’s history from the parents’ perspective and information as to how the person functions in a variety of situations, thus contributing unique information to the diagnosis of autism (Risi et al., 2006). When the ADOS and ADI-R are used in combination, they correlate more highly with the consensus of clinical judgments on autism and ASD than a single instrument.

Gold Standard for Assessment

Use of DSM criteria by an experienced clinician, plus the ADOS and ADI-R, have become the gold standard diagnostic procedure for characterizing individuals with ASD. If knowledge regarding response to intervention is to advance, it is imperative that such a standard be used in research. Yet in a recent AHRQ Comparative Effectiveness Report regarding therapies for children with ASD, the reviewers found that 125 out of 159 intervention studies failed to use or report such a standard in their study (AHRQ, 2011). The great heterogeneity in the population of individuals with ASD, the inadequate description of the participants on the core features of ASD, and the limited description of the level of functioning limit the ability of researchers to examine characteristics of the child that may modify response to treatment.

CORE SYMPTOMS OF AUTISM

Since its inclusion in DSM-III (APA, 1980) autism has been defined in terms of three core dimensions: impaired social interaction; impairments in communication; and restricted repetitive and stereotypical patterns of behavior, interests, and activities. These criteria were further defined and other disorders were added under the umbrella term of pervasive developmental disorders in DSM-IV. The diagnostic criteria for the autistic disorders, Asperger’s syndrome, and PDD-NOS share many features and differ only in the number of criteria in the domains. For example, both autistic disorder and Asperger’s disorder manifest qualitative impairment in social interaction, and restricted repetitive and stereotyped patterns of behavior, interests, and activities. In order for a diagnosis of autistic disorder to be applied, there must also be delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) social interaction, (2) language as used in social communication, or (3) symbolic or imaginative play. In Asperger’s disorder, there are no clinically significant delays in cognitive development or in the development of age-appropriate self-help skills, adaptive behavior (other than in social interaction), and curiosity about the environment in childhood (APA, 2000).

Differentiating between autistic disorder and Asperger’s disorder with regard to the triad of the core features of autism is one of the concerns that has been raised regarding the adequacy of our knowledge on the natural history of autism (First, 2008). It was felt that the DSM-IV criteria provided adequate information regarding how to evaluate the presentation in preschool and school-age children, but that the items do not adequately apply to toddlers, adolescents, and adults (First, 2008). Marans, Rubin, and Laurent (2005) provide detailed descriptions of core social communication challenges and emotional regulation challenges for individuals with Asperger’s disorder or high-functioning autism for whom receptive and expressive language and cognitive level are in at least the average
range. These challenges are presented in Table 1.1 as more detailed examples of the social communication criteria.

New Approach—Two Dimensions

In *DSM-V*, the core features have been reduced to two dimensions: (1) social communication and (2) restricted interests and repetitive behavior. One can find many individuals with an autism diagnosis who are highly verbal, with extensive vocabularies, but who do not understand the pragmatics of language. Pragmatics includes the skills that permit us to understand nonverbal communication, take another’s perspective, recognize and interpret emotions, and understand jokes and double meanings.

The social communication dimension has three subdomains: (1) deficits in social–emotional reciprocity; (2) deficits in nonverbal communication behaviors used for social interaction; and (3) deficits in developing and maintaining relationships, appropriate to developmental level. Each subdomain includes a number of criteria, and an individual will need to meet criteria in all three subdomains to be diagnosed with an ASD when *DSM-V* is applied.

Within the second domain of RRB, there are four subdomains. They are: (1) stereotypical or repetitive speech, motor movements, or use of objects; (2) excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior, or excessive resistance to change; (3) highly restrictive, fixated interests that are abnormal in intensity or focus; and (4) hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of environment. If a

<table>
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<tr>
<th>TABLE 1.1</th>
<th>Core Social Communication Challenges in High-Functioning Autism and Asperger’s Disorder</th>
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<tbody>
<tr>
<td><strong>Capacity for joint attention</strong></td>
<td><strong>Capacity for symbol use</strong></td>
</tr>
<tr>
<td>1. Understanding the communicative intentions and emotional state of a social partner</td>
<td>1. Acquiring higher-level linguistic rules, grammar and syntax, that clarify one’s intent (e.g., subordinate clauses and conjunctions) across social partners and environments</td>
</tr>
<tr>
<td>2. Interpreting and using nonverbal communicative signals (e.g., facial expressions, prosody, body orientation and proximity, and gestures) as they relate to one’s attentional focus, affective state, and intentions</td>
<td>2. Understanding and using verbal conventions for initiating, exchanging turns, and terminating interactions across different social partners and social situations (e.g., rules of politeness)</td>
</tr>
<tr>
<td>3. Considering appropriate topics of conversation, maintaining information, sharing across turns, and repairing communicative breakdowns based on the social context and a listener’s perspective</td>
<td>3. Interpreting and using language in a flexible manner by responding to language that may contain: multiple meaning words, nonliteral language, and irony</td>
</tr>
<tr>
<td>4. Modifying interpretation of more ambiguous language forms (e.g., sarcasm, humor, figurative expressions, etc.) depending upon the intentions or perspective of one’s social partner</td>
<td>4. Using language as a tool for emotional regulation (e.g., preparing for changes in routine, preparing for the expectations of different social contexts, and using appropriate means to request assistance and comfort across social settings and social partners)</td>
</tr>
</tbody>
</table>

Adapted from Volkmar et al. (2005), p. 979.
person meets the behavioral criteria on at least two of the four subdomains of RRB, they qualify for a diagnosis of ASD.

Patterns and Age of Onset

One of the defining features of ASD is age of onset. Kanner (1943) argued that autism was congenital. Rutter (1978) proposed the criterion of onset before 2.5 years of age. Interest in the issue of age of onset has been investigated from multiple perspectives. Three patterns for the emergence of symptoms are described in the literature (Ozonoff, Heung, Byrd, Hansen, Hertz-Picciotto, 2008). The first pattern is that of identification of symptoms in the first year of life, labeled early-onset pattern. The second pattern, termed developmental or autistic regression, is defined as a loss of previously acquired social, communication, and motor skills prior to 36 months of age, after “near-typical” development. The third pattern involves that of near-typical milestone achievement up to 2 years, which is followed by a plateau of skill development.

Researchers have looked at these patterns in terms of the prevalence of each pattern and in terms of the relationship to child outcomes. Kokoyachi and Murata (1998; as cited in Kalb, Law, Landa, & Law, 2010) estimate the range of prevalence of the regression pattern extends from as high as 50% to a low of 15% to 18% when a strict definition of regression is used. With regard to outcomes, the data are mixed, with some studies reporting poorer outcomes in relationship to regression and others reporting minimal to no differences in outcomes (Kalb et al., 2010).

Developmental Characteristics and Outcomes

Contrary to expectations, children who were diagnosed at an early age were not found to be more severely affected than those who were diagnosed later (Landa, Holman, & Garrett-Mayer, 2007; Werner, Dawson, Munson, & Osterling, 2005). Kalb and colleagues (2010) investigated relationships among developmental characteristics and outcomes in a large sample of children assigned to groups based on the three patterns of onset of symptoms. The data for the study came from the Interactive Autism Network (IAN). Families voluntarily submit their data to the IAN registry (http://www.ianproject.org), which began in 2007 and now has data for more than 10,000 children. The sample for this study included 2,720 children of 3 to 17 years of age. Eighty three percent of the sample was male and 90% was white. Of the three patterns, 44% showed skill loss, 39% showed no loss, and 17% showed a plateau. Kalb and colleagues (2010) found that the children with regression showed milder delays in early development, but were at increased risk for poorer outcomes and more likely to have elevated autism symptom scores, which means they were more likely to have a diagnosis of autistic disorder, as opposed to Asperger’s disorder or PDD-NOS.

In an effort to correct for inherent problems in parent recall regarding patterns of onset, Ozonoff and colleagues (2010) examined videotapes for the emergence of behavioral signs of autism in infants in two groups: low and high risk for autism. This longitudinal, prospective study was accomplished by rating videotapes of these children at 6, 12, 18, 24, and 36 months of age. Children were diagnosed as either ASD or typically developing by 36 months of age. The specific behaviors the researchers looked at included the frequency of gaze to face, gaze to objects, smile, nonverbal vocalizations, and word and phrase verbalizations. Raters of the videotapes were blind to child classification. The authors found no differences between the groups on the rate of the three counts of social communication behaviors, nor on the global ratings at 6 months. By 12 months, the groups differed on gaze to face and directed
vocalizations. At 18, 24, and 36 months, there were significant differences between the groups on all four measures. Both groups showed linear patterns of growth, but the trajectory for the children with autism was significantly slower.

These findings have implications for both the diagnosis and the treatment of autism. Ozonoff and colleagues (2010) argued that the defined categories of early onset, regressive autism, and plateau do not accurately portray how symptoms of autism emerge and that specific probes into social development at 6-month intervals are more useful. The high correlation between the global ratings and the specific probes suggest a fruitful area for development of a screening measure. Finally, Ozonoff and colleagues (2010) suggested that the current characterization of types of onset are not useful in terms of predicting outcomes and are problematic with respect to clarity of definition and recall bias. They proposed that a more useful conceptualization is a time-based continuum. At one end are children whose loss of social interest is early, that is, between 6 and 12 months in onset. At the other end of this continuum are children whose loss of social interest and communication skills are so late that “regression” appears quite dramatic (Ozonoff et al., 2010). They further argue that identification of autism by the first birthday may not be possible in the majority of affected children. Furthermore, they argue that in addition to the recommendation for screening at 18 and 24 months, screening may need to occur later to identify these late-onset children. Finally, they make a point that any infant or toddler who demonstrates a sustained reduction in social responsiveness is a candidate for focused intervention to address social communication, whether or not the child screens positive for autism.

Severity of Presentation of Symptoms

One of the limitations of the earlier versions of the DSM is that the criteria are framed as simply present or absent, therefore not allowing for a systematic judgment regarding the severity of the expression of the condition. In the DSM-IV, severity was reflected by whether or not a person was diagnosed with autistic disorder or with the “broader phenotype” of high-functioning autism, Asperger’s disorder, or PDD-NOS. In the DSM-V criteria, it is proposed that each of the two dimensions—deficits in social communication and restricted repetitive behavior—be rated for severity. Table 1.2 contains the proposed statements for each of these levels of severity ranging from: (1) normal variation; (2) substantial symptoms; (3) requires support; (4) requires substantial support; and (5) requires very substantial support. It is expected that this proposed system of classifying severity will contribute to a characterization of people’s symptoms and a better basis for planning intervention (Lord, 2011).

Medical Diagnosis or Educational Identification

One controversy in the field of ASD is the debate as to whether or not a medical diagnosis is necessary for educational programming. Some argue that given that ASD is defined in terms of specific behaviors or absence of behaviors, identification of educational needs without an actual diagnosis is adequate. One of the potential sources of data regarding the prevalence of ASD is the annual child count data. In an educational identification context, the condition must have a “significant impact on the child’s educational, emotional, or functional skills, such that specialized instruction is necessary for the child to be successful. In Colorado the official position of the Colorado Department of Education is that a medical or clinical diagnosis does not automatically qualify for special education” (Colorado Department of Education, 2008).
There is considerable anecdotal evidence that schools are reluctant to identify students as having an autism educational exceptionality due to the considerable expense likely to be involved if a family demands intensive treatments using a particular methodology. However, the Individuals with Disabilities Education Act (IDEA; 1990) based definition of an educational exceptionality on the impact of educational, emotional, or functional skills. Too often we have encountered situations in which the child’s speech is at age and grade level, but the pragmatics of communication are missing. Thus, the child may well meet many of the DSM criteria for a diagnosis on the autism spectrum, but because his or her academic work is at age level, the child is not seen as eligible for educational intervention.

There is also reason to believe that there are a number of children on the autism spectrum who are in the general education classes without a classification and therefore without an individualized educational plan (IEP). Hepburn and colleagues (2008) compared a teacher nomination strategy using a six-question checklist to the performance of the Autism Spectrum Screening Questionnaire (ASSQ; Ehlers, Gillberg, & Wing, 1999) as a strategy for identifying children with an ASD. Teachers were asked to nominate up to two children in their general education elementary classrooms (K–5) who met any of the six characteristics listed in Exhibit 1.1.

A total of 60 teachers with 1,355 students among them participated. Fifty of the 60 teachers nominated at least one child, with a total of 116 children nominated. Ninety-five children scored 17 or higher on the ASSQ, with 9.2% of boys and 5% of girls receiving scores of 17 or higher. Agreement between being nominated and scoring 17 or higher on the ASSQ was 93% to 95%. Of those nominated, 32% currently had an IEP, previously had an IEP, or had an IEP in process, and 68% were not identified as a special education student. The nomination strategy seems to have promise as a more time- and cost-efficient first-level screener for children who need assistance with the social aspects of school.

Marans, Rubin, and Laurent (2005) identified social communication skills as a critical priority for educational programming, and yet these skills, or lack thereof, are not adequately identified and attended to in educational programs. The greatest challenges in social communication for these children lies ahead, as they enter elementary, middle, and high school

### TABLE 1.2

<table>
<thead>
<tr>
<th>Proposed dimensional ratings for ASD in DSM-V</th>
<th>Social communication</th>
<th>Fixated interests and repetitive behaviors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Requires very substantial support</td>
<td>Minimal social communication</td>
<td>Marked interference in daily life</td>
</tr>
<tr>
<td>Requires substantial support</td>
<td>Marked deficits with limited initiations and reduced or atypical responses</td>
<td>Obvious to the casual observer and occur across context</td>
</tr>
<tr>
<td>Requires support</td>
<td>Without support, some significant deficits in social communication</td>
<td>Significant interference in at least one context</td>
</tr>
<tr>
<td>Subclinical symptoms</td>
<td>Some symptoms in this or both domains; no significant impairment</td>
<td>Unusual or excessive but no interference</td>
</tr>
<tr>
<td>Normal variation</td>
<td>May be awkward or isolated but within normal limits (WNL)</td>
<td>WNL for developmental level and no interference</td>
</tr>
</tbody>
</table>

Adapted from Lord (2011).
and encounter much more complex versions of joint attention skills, mastering of higher-level linguistic rules, and use of language and cognition for self-regulation and mutual regulation, (Marans et al., 2005). In theory, identification of “educationally significant needs” should be adequate to address the needs of students and a medical diagnosis of ASD should not be necessary for appropriate educational intervention. In practice, we have found reason to question such an assumption.

CO-OCCURRING CONDITIONS FOR CHILDREN WITH ASD

People with ASD may have developmental, neurological, medical, and psychiatric conditions in addition to ASD. Sometimes these conditions are referred to as comorbid conditions (Matson, LoVullo, Rivet, & Boisjoli, 2009; Volkmar and Klin, 2005) and sometimes as co-occurring or associated conditions (Levy, Giarelli, Lee, Schieve, Kriby, Cunniff, et al., 2010; Reaven, 2009). Levy and colleagues (2010) argued that the term comorbid may imply a condition that is distinct from the disorder itself or may imply causality. For this reason, the authors prefer the term co-occurring condition.

LITERATURE ON CO-OCCURRING CONDITIONS

Most of the literature documenting co-occurring conditions is based upon clinical samples and can have widely varying figures regarding prevalence, presumably due, in part, to the type of practice and the source of the sample. Levy and colleagues (2010) used population-based data to describe four groupings of co-occurring disorders. They are: (1) developmental diagnoses, (2) psychiatric, (3) neurological diagnoses, and (4) possibly causative medical diagnosis (see Table 1.3). Levy and colleagues (2010) also found that 60% of the sample of 2,568 children who meet Autism and Developmental Disabilities Monitoring (ADDM) network criteria for a case definition had one co-occurring diagnosis, while 26% had two or more diagnoses or symptoms consistent with one of the four types of conditions for a total of 86% with a co-occurring disorder. In addition, they reported that those children who were diagnosed with ASD later had a greater number of preceding co-occurring diagnoses. The authors postulated that other conditions may have a masking effect in the diagnosis of ASD. This data came from 8-year-old children born in 1994, and ADDM surveillance has continued with

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<tr>
<td><strong>QUICK SCREENING QUESTIONS ON ASD CHARACTERISTICS</strong></td>
</tr>
<tr>
<td>1. Is the person socially awkward?</td>
</tr>
<tr>
<td>2. Does the person not seem to understand the feelings of others?</td>
</tr>
<tr>
<td>3. Does the person talk a lot about own interests, but not very good at conversations?</td>
</tr>
<tr>
<td>4. Does the person not chat with peers just to be friendly?</td>
</tr>
<tr>
<td>5. Does the person demonstrate a lack of flexibility?</td>
</tr>
<tr>
<td>6. Does the person demonstrates intense interest in just a few topics or activities?</td>
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EXHIBIT 1.1

QUICK SCREENING QUESTIONS ON ASD CHARACTERISTICS

<p>| 1. Is the person socially awkward? |
| 2. Does the person not seem to understand the feelings of others? |
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| 5. Does the person demonstrate a lack of flexibility? |
| 6. Does the person demonstrates intense interest in just a few topics or activities? |</p>
<table>
<thead>
<tr>
<th>Developmental diagnoses</th>
<th>n (%)</th>
<th>Psychiatric diagnoses</th>
<th>n (%)</th>
<th>Neurological diagnoses</th>
<th>n (%)</th>
<th>Possibly causative medical diagnoses</th>
<th>n (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>All cases developmental</td>
<td>2,123 (82.7%)</td>
<td>All cases psychiatric</td>
<td>258 (10.0%)</td>
<td>All cases neurological</td>
<td>404 (15.7%)</td>
<td>All cases possible causative diagnosis</td>
<td>95 (3.7%)</td>
</tr>
<tr>
<td>diagnosis</td>
<td></td>
<td>diagnosis</td>
<td></td>
<td>diagnosis</td>
<td></td>
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<td></td>
</tr>
<tr>
<td>Specific developmental</td>
<td>%</td>
<td>Specific psychiatric</td>
<td>%</td>
<td>Specific neurological</td>
<td>%</td>
<td>Specific causative</td>
<td>%</td>
</tr>
<tr>
<td>diagnosis</td>
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<td></td>
<td>diagnosis</td>
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<td>diagnosis</td>
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<tr>
<td>Language disorder</td>
<td>63.4</td>
<td>ODD</td>
<td>4.0</td>
<td>Epilepsy</td>
<td>15.5</td>
<td>Other genetic/congenital</td>
<td>1.0</td>
</tr>
<tr>
<td>ADHD</td>
<td>21.3</td>
<td>Anxiety disorder</td>
<td>3.4</td>
<td>Encephalopathy</td>
<td>5.9</td>
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<td></td>
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<tr>
<td>Intellectual disability</td>
<td>18.3</td>
<td>Emotional disorder</td>
<td>2.4</td>
<td>Hearing loss</td>
<td>1.7</td>
<td>Down syndrome</td>
<td>0.8</td>
</tr>
<tr>
<td>Sensory integration</td>
<td>15.7</td>
<td>Mood disorder</td>
<td>2.3</td>
<td>Cerebral palsy</td>
<td>1.7</td>
<td>Chromosome disorders</td>
<td>0.5</td>
</tr>
<tr>
<td>Learning disorder</td>
<td>6.3</td>
<td>OCD</td>
<td>2.0</td>
<td>Visual impairment</td>
<td>1.0</td>
<td>Fragile X syndrome</td>
<td>0.3</td>
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<tr>
<td></td>
<td></td>
<td>Depression</td>
<td>1.1</td>
<td>TS/tics</td>
<td>0.5</td>
<td>Tuberous sclerosis</td>
<td>0.2</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Bipolar disorder</td>
<td>0.7</td>
<td>Brain injury</td>
<td>0.4</td>
<td></td>
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<tr>
<td></td>
<td></td>
<td>Mutism</td>
<td>0.5</td>
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<tr>
<td></td>
<td></td>
<td>Psychosis</td>
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<td></td>
<td></td>
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<tr>
<td></td>
<td></td>
<td>RAD</td>
<td>0.3</td>
<td></td>
<td></td>
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</tr>
<tr>
<td></td>
<td></td>
<td>Conduct disorder</td>
<td>0.2</td>
<td></td>
<td></td>
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</tr>
<tr>
<td></td>
<td></td>
<td>Schizophrenia</td>
<td>0.1</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Diagnoses are not mutually exclusive (e.g., one subject may have more than one diagnosis).

Genetic or congenital syndromes or disorders (e.g., embryopathy, fetal alcohol syndrome, Cornelia De Lange syndrome, and others).

Chromosomal disorders (e.g., deletion, duplication, and others). ODD, oppositional defiant disorder; OCD, obsessive–compulsive disorder; RAD, reactive attachment disorder; TS, Tourette syndrome; VCF, velocardiofacial syndrome.

Source: Reprinted with permission from Levy et al. (2010).
new birth cohorts in 4-year increments. Similar analyses with subsequent ADDM birthcohorts will provide information as to the reliability of these findings.

While coping with the diagnosis of an ASD is difficult in and of itself, the presence ofone or more co-occurring conditions can complicate day-to-day care. The proposedDSM-V guidelines include an appraisal of all four categories noted above as essential to therecommended comprehensive approach to assessment. The DSM-V approach looks at thediagnosis of ASD as a factor in and of itself, with the expectation that all co-occurring condi-tions should be evaluated as well, not for the purpose of diagnosis, but for the purpose ofprescribing effective interventions.

Co-occurring Psychiatric Disorders

As previously noted, use of the term co-occurring condition avoids any assumptions ofunderlying etiology for the condition. Specifically, the position is that an individual who meets diagnostic criteria for a psychiatric condition by definition has that condition. Recog-nition of both conditions as having independent diagnostic criteria has important impli-cations for access to care, such that different treatments and follow-up care may be required.

Within the DSM system ASD and psychiatric diagnoses are Axis I disorders. This factprobably contributes to the reluctance of some clinicians to apply both an ASD diagnosis and an Axis I psychiatric diagnosis to the same person. Some argue that if a person’s anxiety disorder, for example, is due to the fact that the person has autism, then from that perspective, the anxiety disorder is not amenable to treatment from the mental health system. In this example, we see the potential negative consequences of attributing symptoms to a presumed etiology. The person may not receive appropriate treatment.

THE CASE OF JAMES

Reaven (2009) offered an example of James, an 8-year-old boy with a diagnosis of Asperger’sdisorder with intellectual and academic abilities in the average range. His IEP was designed toaddress the ASD core symptoms of problems with social communication, independence, andorganization. At the end of second grade, he began to express fear of making mistakes, and bythird grade, his worries about making mistakes increased and he frequently asked to stayhome from school. He was also bothered by loud noises to the extent that he met clinical cri-teria for generalized anxiety disorder and specific phobia. Reaven (2009) went on to describehow a cognitive behavioral therapy (CBT) approach can be successfully modified for use forchildren with ASD. Reaven (2009) recommended a variety of strategies, including extensiveuse of concrete supports such as video modeling and using the individual’s specific interests or talents as a platform to build upon. The modification of CBT developed by Reaven and hercolleagues is now available in a book, Face Your Fears (Reaven, Blakeley-Smith, Nichols, &Hepburn, 2011). Increasingly, investigators are developing diagnostic and treatment strat-egies for co-occurring psychiatric conditions for individuals with an ASD (Chalfant, Ropee,& Carroll, 2007; Sofronoff, Attwood, & Hinton, 2005; Sze & Wood, 2007).

Co-occurring Medical Conditions

In order to provide a complete assessment of the individual with ASD, it is important to lookat all associated medical problems the individual may have which are relevant to his or hercomprehensive clinical management (APA, 2000). These problems should be identified
under Axis III of the complete clinical workup. In addition to specific neurological conditions, such as seizure disorders, there are a number of other medical problems often overlooked or undiagnosed in people with ASD. Nurses and physicians who care for people with ASD and their families have frequently cited the difficulty children have with sleep, adequate nutrition, gastrointestinal disturbance, food selectivity, and self-injury, to name just a few. Sleeping disorders and selectivity and rigidity in eating patterns can be grave issues for many families. These issues may not be satisfactorily diagnosed or treated, when in fact, treatment of such issues as sleep deprivation and hunger could ameliorate irritability, inability to concentrate, and other symptoms that intersect with those specific to ASD.

Access to medical care for co-occurring conditions can be difficult for many families. In some cases, primary-care providers may not feel capable of managing such complex problems. Parents may seek the help of a developmental behavioral pediatrician to work with their pediatric primary-care providers, but collaborative consultation can be difficult to realize. The Autism Treatment Network (ATN) consists of 17 centers, each with some funding from Autism Speaks and the Health Resource Service Administration (HRSA) under the Combating Autism Act. These centers each have a common purpose of developing our knowledge about the prevalence and range of co-occurring medical problems and assessing methods for managing these conditions (ATN, 2011). The ATN has also established a data registry composed of systematically collected and consistent information from families about common co-occurring medical conditions.

Co-occurring ASD and Specific Disorders or Syndromes

In the *Handbook of Autism and Pervasive Developmental Disorders*, Filipek (2005) makes the point that the relationship between ASD and medical conditions can be viewed from two approaches: as medical conditions associated with ASD or as ASD associated with medical conditions. One can look for conditions in a population of people with ASD or one can look for ASD in a population of individuals with a given syndrome. Some may argue “why should we have another label?” The counter-argument is that for the child who has Down syndrome (DS) and ASD, for example, it is not typical for a child with DS to not be talking, while it is an all too frequent problem for a child with autism. What is known about communication for children with ASD should inform the treatment plan for the child with DS.

Regarding the prevalence of ASD among children with DS, DiGuiseppi and colleagues (2010) recruited children with DS from a birth registry sample. All families still residing within a 10-county area, in which the children were still alive, were invited to participate in a study to look at their child’s social, communication, and behavioral needs. Depending upon the age of the child, the Modified Checklist for Autism in Toddlers (M-CHAT; Robins & Dumont-Mathieu, 2001), or the Social Communication Questionnaire (SCQ; Rutter, Bailey, & Lord, 2003) was administered in a telephone interview. All of the families of children who screened positive were invited to come in for an evaluation that included the ADOS (Lord et al., 1999), ADI-R (Rutter, LeCouteur, & Lord, 2003), an age-appropriate developmental assessment, and the Vineland Adaptive Behavior Scales, Second Edition (Sparrow, Balla, & Cicchetti, 1984). In this sample, the estimated prevalence of ASD (broad phenotype) was 18.2% and that of autistic disorder was 6.4%. This estimate indicates that ASD is 17 to 20 times higher among children with DS than the estimated prevalence of ASD in the general population.

Down syndrome is just one example of a genetic syndrome that appears to have a higher than expected co-occurrence with ASD when compared to the general population (DiGuiseppi
et al., 2010). Studies such as this give credence to the concerns of families with a child with a given syndrome that presents with atypical characteristics. In response to this issue, groups such as the Colorado Down Syndrome-Autism Connection have been formed (Zaborek, 2011). These families have come together in a support group in which they feel more comfortable and provide advocacy for one another.

**Autism and Co-occurrence with Intellectual Disability**

In Kanner’s original description of infantile autism, he made a point of distinguishing autism from mental retardation (Schalock et al., 2010). In the 1940s, and for some time thereafter, the predominant view was that mental retardation was not amendable to intervention. At that time, there was not a uniformly accepted definition of mental retardation. In 1961, the American Association on Mental Deficiency (AAMD), now the American Association of Intellectual and Developmental Disabilities, proposed a definition of mental retardation that soon became the standard (Heber, 1961). This definition is relevant here due to its emphasis on measured intelligence and measured adaptive behavior, and because an underlying etiology was not part of the definition. Important elements of this definition are that the individual’s scores on a norm-referenced standardized test of intelligence and test of adaptive behavior are below 70 and that these problems are apparent before 18 years of age. Once the AAMD definition became commonly adopted, the issue of etiology became irrelevant to the diagnosis of mental retardation.

In 2006, Edelson published a review of the literature on the co-occurrence of autism and mental retardation. She reviewed all reports she could find that spoke to both autism and mental retardation from 1943 through 2003, including books and book chapters, as well as journal articles. In her review, Edelson (2006) examined three questions, each of which was important to understanding the diagnosis of ASD and mental retardation. Her three questions were: (1) Do the prevalence rates (of co-occurrence of mental retardation) reported in the literature derive from empirical sources? (2) Can nonempirical sources of these statistics be traced historically to valid empirical studies? (3) When empirical studies have been conducted, are the methods by which intelligence is assessed appropriate? For the time period from 1943 to 2003, she found 145 articles that met at least one of the following three criteria: (1) level of intelligence in children with autism was investigated; (2) cognitive abilities of children with autism were discussed; or (3) cited a claim about the co-occurrence between autism and mental retardation. She found that only 26% of the claims about prevalence of mental retardation among people with ASD were derived from empirical studies. From the 53 empirical articles that spanned almost 50 years, she found an average prevalence rate of 75.20%. The lowest prevalence rate, 34.33%, was reported in three articles published prior to 1950. The highest prevalence rate, 86.78%, was reported from 9 studies conducted between 1970 and 1979. Edelson (2006) classified 165 articles as nonempirical in her total set of 215 articles. Only 25% of the nonempirical articles based their claims on empirical studies. Once she traced citations back, she found that the majority of citations were not reported accurately. She made a compelling case that the commonly quoted figure of 70% to 80% was high and not based on population-based studies.

Further compounding the potential for error in the claim that 70% to 80% of children with ASD have mental retardation, is the fact that 55% of the empirical studies were conducted prior to 1980 and 75% prior to 1990. Edelson (2006) pointed out that the claims that a majority of children with autism have mental retardation may be erroneously
referred in journal articles, child psychopathology textbooks, in abnormal psychology textbooks, and most troublesome, in the *DSM-IV-TR* criteria (Edelson, 2006).

In 2007, population-based surveillance data on ASD and associated symptoms became available from the CDC Autism and Developmental Disability Monitoring (ADDM) Network (CDC, 2009). The ADDM results for six states, published in 2009, on children who were 8 years of age in 2006 indicated that the average percent of cases of ASD with IQs below 70 was 41%, with a range of 29.3% to 51.2%. One of the cautions about the ADDM data is that it does not involve direct assessments of the children. ADDM data, however, is the source for the ubiquitous citation of the prevalence of autism as 1 in 110 children. If the field is going to accept the overall prevalence figure for autism in ADDM, then serious consideration needs to be given to the data on associated features, including the figure regarding co-occurrence of mental retardation.

Perceptions have changed regarding the previously held belief that a majority of children with autism have ID (Lord, 2011; Lord & Bishop, 2010). This change resulted in part from a review of the CDC surveillance data, and in part from the new perspective in the *DSM-V* that ID is a separate diagnosis.

**SUMMARY**

Autism spectrum disorder is a complex neurodevelopmental disorder with a range in expression from mild difficulties in social communication to profound difficulties with all aspects of daily living. Autism, by definition, has an onset before 3 years of age and has three core behavioral domains. Since the initial description by Kanner (1943), the criteria for a diagnosis of autism have been refined and broadened to include what is now referred to as the “broader phenotype.” This broader phenotype includes people with high-functioning autism or Asperger’s disorder. Autistic disorder was first included in the *DSM-III* in 1980. At that time, the superordinate category of pervasive development disorder was introduced with the subcategories of autistic disorder, Asperger’s disorder, PDD-NOS, childhood disintegrative disorder, and Rett disorder.

Over the course of revisions of the *DSM* there have been refinements to the criteria for a clinical diagnosis, and on the horizon is *DSM-V*, with adoption expected in 2013. *DSM-V* includes some major changes, each of which reflects research that has been happening over the past 30 years. The recommended term will be ASD with no differentiation within the spectrum. *DSM-V* reduces the core symptom domains from three to two, with the social and communication domains combined into one domain.

Psychometric instruments are available to assist clinicians in diagnosing ASD. These tools, particularly the ADOS and the ADI-R, combined with application of the *DSM* criteria have become the gold standard for making the diagnosis of autistic disorder or the broader phenotype for research purposes. However, much of the work to date has not conformed with this standard, and studies frequently are reported in which there is inadequate detail about the characterization of the subjects (AHRQ, 2011).

From a service perspective, greater support and services are made available when a formal diagnosis of ASD is made. Therefore, clinicians may feel pressured to make a diagnosis of ASD, even when not certain. Children and adults with ASD also have a significant degree of co-occurring developmental, psychiatric, and medical problems. These problems frequently are “overshadowed” by a diagnosis of autism and are not treated as separate conditions. The reverse is also true, where other diagnoses may delay the diagnosis of an ASD. Regardless of the number of co-occurring conditions that an individual may have, each one warrants
thorough assessment. Each individual should receive a comprehensive and developmentally appropriate course of treatment.

REFERENCES


