What Is Autism Spectrum Disorder?

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Abstract

Autism spectrum disorder (ASD) is a neurodevelopmental disorder with social-communication deficits and restricted and/or repetitive behaviors and/or interests. The diagnostic criteria of the disorder have evolved over the years with new research on the features, associated symptoms, prevalence, and etiology of the disorder. This chapter offers an overview of the presentation, development, history, prevalence, and impact of ASD on the child and family. Research on the etiology of ASD, including potential risk factors and dispelled myths, is summarized.

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1.1 The Diagnosis and Presentation of Autism Spectrum Disorder

Autism spectrum disorder (ASD) is a neurodevelopmental disorder associated with deficiencies or excesses in two domains: social-communication and restricted, repetitive behaviors and interests (American Psychiatric Association [APA], 2013). Social-communicative skills and restricted and repetitive behaviors and interests vary across individuals with and without a diagnosis of ASD. These distinct domains can be atypical or normative depending on where an individual falls within the spectrum of the behavior. Behaviors of individuals with ASD and normative samples are etiologically and qualitatively related; however, individuals who do not meet the criteria for ASD may not demonstrate abnormalities in those domains, may exhibit abnormalities in a single domain, or may display minimal difficulties in both domains (Constantino & Todd, 2003). Individuals with ASD must exhibit impairment in social-communication and restricted, repetitive behavior and interest, but they are heterogeneous in presentation and severity of impairment. The purpose of this chapter is to describe the history and presentation of ASD by introducing the diagnostic criteria, common presentation and development of the disorder and comorbidities in children, and risk factors that contribute to the disorder.

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1.1.1 Diagnostic Criteria

The diagnostic criteria for ASD that are most commonly used by clinicians in the United States are derived from the American Psychiatric Association's Diagnostic Statistical Manual, Fifth Edition (DSM-5 2013). The DSM-5 states that ASD impairments in the domain of social-communication include failure to initiate and/or reciprocate emotional and social exchanges, abnormalities in nonverbal communication behavior and understanding, and/or difficulties forming and sustaining relationships. The DSM-5 criteria for restricted interests and repetitive behavior include the presentation of at least two or more of the following: stereotyped or repetitive movements or speech (e.g., flapping arms back and forth or repeating the same sentence/phrase), rigidity in routine, abnormalities in domain or intensity of interests, and/or abnormalities in reactivity to sensory input (APA, 2013). Despite these specific diagnostic criteria, consistency in presentation across and within individuals and reliability of diagnosis are fairly low (Falkmer, Anderson, Falkmer, & Horlin, 2013) depending on developmental period, severity of impairment, and genetic, medical, and psychosocial comorbidities, which are described below. See Table 1.1 for diagnostic criteria and examples.

1.1.1.1 Social-Communication Deficits

Social-communication deficits or excesses are often the first sign of ASD, and can appear within the first year of a child's life (Guthrie, Swineford, Nottke, & Wetherby, 2013; Richler et al., 2006; Sacrey et al., 2015). Early social-communication difficulties may include abnormalities in the use of nonverbal expressive and receptive communication, such as gestures and imitation of facial expressions. Before children can speak, most neurotypical children try to communicate with caregivers by pointing or reaching for things. When neurotypical infants see an object of interest, they may engage in joint attention by looking to the object, then the caregiver, and then back at the object, as if to direct their caregiver's attention to the item of interest (Baron-Cohen, Leslie, & Frith, 1985). When their caregiver points or looks at something, the infant likely follows the direction of the point. Similarly, when the caregiver smiles, the infant likely reciprocates the behavior and smiles back.

For children with ASD, however, many of those social and communicative behaviors are atypical or absent. Many children with ASD do not engage their parents in acts of joint attention, and may not attempt to gain a caregiver's attention (Charman, 2003; Macari et al., 2012), for example, by pointing or gesturing (Macari et al., 2012). Additionally, some children with ASD lack imitation skills (see review in Jones, Gliga, Bedford, Charman, & Johnson, 2014). For example, if a parent shakes a rattle or puts blocks together, a child with ASD may not imitate those behaviors. Other atypical behaviors include avoiding looking at faces, glancing at a face quickly, or focusing on parts of the face that do not communicate emotions (Jones et al., 2014). Because infants learn language, communication, and social behaviors through joint attention and imitation (e.g., Charman, 2003), infants and young children with deficits in these areas may miss valuable learning opportunities, which may contribute to more significant and more pronounced impairments at a later age (Dawson, 2008).

As children grow, neurotypical children begin to display interest in and then seek out peers to play with. Some children with ASD seem to avoid social play opportunities, whereas others may desire relationships but do not know how to initiate or maintain them. Such a child may hover on the outskirts of a peer group, but not ever integrate into the group, even when invited to do so. Some children with ASD spend more time in solitary play, even when peers are present (Zager, Cihak, & Stone-MacDonald, 2017), while other children with ASD may attempt to play with peers but do not exhibit the foundational social skills necessary to engage in reciprocal play behavior. For example, a child with ASD may not be skilled in sharing or turn-taking or may not pick up on verbal and nonverbal cues that guide interaction and indicate how a game should be played. A child with ASD may not understand the

Table 1.1 DSM-5 diagnostic criteria and examples

Domain	Diagnostic criteria	Examples
Social-communication Deficits	Failure to initiate and/or reciprocate emotional and social exchanges	• Looks down when someone says, "Hi" • Responds to a peer's description of weekend activities with an off-topic monologue
	Abnormal nonverbal communication behavior and understanding	• Avoids eye contact • Facial expressions and/or tone of voice seem flat or robotic
	Difficulties forming and sustaining relationships	• Plays alone instead of with others • Avoids physical touch
Restricted, repetitive interests, behaviors, and activities (at least two)	Stereotyped or repetitive movements or speech	• Flaps hands repeatedly • Organizes toys instead of playing with them
	Rigidity in routine	• Throws a tantrum when a stop is added on the typical drive home • Insists on looking in every window he or she passes by
	Abnormalities in domain or intensity of interests	• Talks almost exclusively about a collection of old video game consoles • Stares at the wheel of a toy car, instead of the whole car
	Abnormalities in reactivity to sensory input	• Cries when in a place with bright lights or loud noises • Does not show a reaction to a sudden loud noise, like an alarm or clap of thunder

concept of a "do-over" and may become frustrated at the perception that another child is not following the rules. This unawareness or failure to comply with social norms can lead to peer rejection (Schroeder, Cappadocia, Bebko, Pepler, & Weiss, 2014).

Another key skill that most children with ASD lack is often labeled theory of mind (Baron-Cohen et al., 1985). Theory of mind is the ability to perceive or understand other people's perspectives (Wellman, Cross, & Watson, 2001). Children with ASD are typically more concrete and often misinterpret others' behaviors and miss important social cues. For example, children with ASD may not realize that it is inappropriate to enter into a conversation with a group of individuals who are talking to one another in a heated or an animated manner or may make a factual statement about another person that may be hurtful without considering the other person's feelings. Children with ASD may also struggle to understand facial expressions and the cause of others' emotions. For example, a child with ASD may, along with peers, learn that another child in the class was seriously injured. Most peers may cry or otherwise express distress yet the child with ASD may appear unaffected and may even question the behavior of peers, "Why are they crying?" (Bauminger, 2002). Many also struggle to identify, cope with, and appropriately express their own emotional states. For example, some children with ASD may not identify their feeling as "angry" despite yelling, hitting, and clenching their fists (Mazefsky, Borue, Day, & Minshew, 2014).

Additionally, many children with ASD struggle during play due to deficits in imitation, understanding of symbolism (i.e., use of objects, actions, or ideas to represent other objects, actions, or ideas; Prizant, Wetherby, Rubin, & Laurent, 2003), imagination, and social understanding (Bauminger, Shulman, & Agam, 2003). Most preschoolers engage in imaginative and symbolic play, such as pretending to make dinner in a toy kitchen and using the toy stove to "cook." However, a child with ASD who has limited imitation or creative play may not know how to join the play. When children with ASD avoid, learn to avoid, or are rejected from play and social experiences, they miss important modeling and learning opportunities, which may exacerbate their deficits (Dawson, 2008). Children with ASD's social difficulties are further compounded by excessive repetitive and restricted behaviors, interests, and activities, which may also impact their social engagement and opportunities.

1.1.1.2 Repetitive and Restricted Behaviors, Interests, and Activities

Repetitive and restrictive behaviors often become most apparent when a child begins to play with toys independently and develop language. There is some research reporting repetitive and restrictive behaviors in children with ASD by the second year of life (e.g., Wetherby et al., 2004), while other studies report that those behaviors only become atypical later in childhood (e.g., Werner & Dawson, 2005). The presentation, assessment, and treatment of repetitive and restricted behaviors, interests, and activities will be covered in greater depth in Chap. 9, so they are only briefly reviewed here.

Some repetitive and restricted behaviors change as children develop, as interests and skills change. For example, a young child may repeatedly line up blocks or other toys instead of building or playing with them, and then, in later years, begin to insist that his or her clothes be hung in a particular manner and that other precise organizational patterns are followed (Watt, Wetherby, Barber, & Morgan, 2008). Stereotyped behaviors can also appear in the use of language, such as repeating one word or phrase (echolalia) or only repeating information on one topic that is of interest to them, which may also make the individual seem "rigid" (APA, 2000).

Children with ASD can also exhibit rigidity in their adherence to routines, social flexibility, and understanding of rules. For example, some children with ASD have trouble adapting to unexpected changes to schedules. Some may become upset when other children want to invent new games or alter the rules in games because they do not understand that some rules can be flexible (Hobson, Lee, & Hobson, 2008). Children with

ASD may also display rigid and atypical interests such as in the mechanics of toys, rather than the function (Ozonoff et al., 2008). For example, while neurotypical children might roll a toy car on the floor and make car noises such as the noise of a horn, a child with ASD might be more likely to play with a toy car by staring at the spinning wheels, repeatedly opening and shutting the hood, or lining up all the toy cars in a row. Children with ASD may have very restricted interests, such as exclusive focus on batting averages in major league baseball, or in types and functions of different vacuum cleaners. Some children become focused on very specific environmental stimuli, such as a moving ceiling fan or reflections in car windows. Vocal children with ASD may focus most or all conversation on their restricted interests and fail to pick up on signals that their conversational partner has lost interest in the topic.

In addition to stereotyped behavior and restricted interests, many children with ASD have abnormal reactions to sensory stimuli that are considered repetitive and restricted behaviors (APA, 2013). Some children with ASD are hypersensitive to sensory experiences, such as reacting negatively to loud noises, bright lights, strong tastes, or physical touch. In contrast, some children with ASD are hyposensitive to sensory stimuli. This is referred to as sensory under responsivity and often manifests as failure to exhibit discomfort or to communicate pain (Hazen, Stornelli, O'Rourke, Koesterer, & McDougle, 2014). For example, a child with ASD may show no reaction when he or she bangs his or her head on the table yet demonstrate clear indicators of pain when he or she trips and falls. Some children with ASD don't react to even extreme temperatures, such as seeming not to be cold even when the temperature is quite low. Other children may not react to loud and sudden noises, even when the noise was so extreme that everyone around exhibits a startle response. As with other domains, the response to environmental stimuli is variable among children with the same diagnosis.

The range in presentation of DSM-5 criteria alone demonstrates the heterogeneity within the disorder. Deficits in both core domains can affect movement, speech, interests, and reactions, and children with ASD can present with any combination of types or presentations of abnormalities. Further contributing to differences in presentations is a variety of deficits that are commonly associated with ASD diagnoses.

1.1.2 Associated Deficits and Abnormalities

While not part of the diagnostic criteria, children with ASD often exhibit a range of cognitive, linguistic, and adaptive living deficits, as well (Ousley & Cermak, 2014). Deficits in these other domains are not currently included as core deficits in the diagnosis of ASD because it is unclear if they are caused by comorbid disorders, if they overlap with other disorders because the disorders are related, or if they are more central deficits of ASD (Mazefsky et al., 2014). These commonly co-occurring impairments are noteworthy as they affect presentation and treatment.

1.1.2.1 Cognitive Impairments

Both global cognitive functioning and specific cognitive abnormalities are common in children with ASD but there is no singular cognitive profile (Joseph, Tager-Flusberg, & Lord, 2002). Global cognitive ability can range from intellectual impairment to above-average intelligence, as will be discussed more in the section on comorbidities. But, even children with ASD with above-average intelligence often exhibit some specific cognitive deficit. Common cognitive abnormalities in this population include deficits in executive functioning, a bias towards details instead of the larger picture, the ability to process large amounts of information, cognitive flexibility, and learning and processing speed (DeMyer, Hingtgen, & Jackson, 1981; Minshew & Williams, 2007). Deficits in executive functioning will be reviewed more in the section on comorbidities because they often result in a diagnosis of attention-deficit hyperactivity impulsivity (ADHD) disorder, but the deficits may include problems with working memory and the ability to

inhibit impulses, organize, plan, and execute strategies (Ozonoff & Stayer, 2001). All of these problems can make it difficult for children with ASD to organize large amounts of information together or to break large amounts of information down into manageable parts.

Relatedly, a bias towards focusing on details may make it hard for the child to take a broad perspective or to learn and process large amounts of information (Happé & Frith, 2006). Some children with ASD exhibit superior processing of details, such as the ability to detect modifications to melodies in music (Mottron, Peretz, & Ménard, 2000) or faster performance on spatial tasks, like map learning, because they have a preference for processing details (Caron, Mottron, Rainville, & Chouinard, 2004). The preference for details can be a strength that helps the child excel in fields that value details, like mathematics, engineering, or music. The processing bias can also detract from the child's perception of the larger picture, in some instances. It remains unclear if these children with ASD have true deficits in global processing, or if their global processing is just negatively impacted by the focus on details sometimes, but bias towards details should be considered as it can affect the child's social, emotional, and cognitive behaviors (Happé & Frith, 2006). The focus on details is not present in all children with ASD and given the heterogeneity in cognitive ability within and between children with ASD in all cognitive domains it is important to assess each individual's relative strengths and weaknesses to ascertain where they may excel and where they may need additional support.

1.1.2.2 Linguistic Deficits

Many children with ASD also need support and early intervention due to linguistic deficits beyond social-communication abnormalities (Kim, Paul, Tager-Flushberg, & Lord, 2014). A majority of children with ASD develop expressive and receptive language (Norrelgen et al., 2014), but they do so later and at a slower rate than neurotypical children do (Kim et al., 2014). Some children with ASD have relatively normal language development but make grammatical errors or exhibit abnormalities in prosody (speech rhythm, stress, and intonation; Charman, Drew, Baird, & Baird, 2003; Eigsti, de Marchena, Schuh, & Kelley, 2011). Finally, some children with ASD do not develop spoken communication or phrase speech at all (Kim et al., 2014; Norrelgen et al., 2014).

For children with ASD who do develop spoken communication, they may exhibit deficits in expressive language, receptive language, or both. Early signs of deficits and delays in receptive language include failure to respond to the sound of one's name (Nadig et al., 2007) or a mother's voice in infancy (Klin, 1991), and lack of understanding of instructions at an older age. Expressive language delays include a delayed average age of first word production; the average is 38 months for children with lower functioning ASD, compared to an average age of 8–14 months for neurotypical children (Howlin, 2003). Additionally, some toddlers and children with ASD produce noises that are inappropriate in content, volume, or clarity and some exhibit echolalia, or repetition of others' words, phrases, and/or intonation (Kim et al., 2014).

Other linguistic errors and oddities can be seen in children with ASD and language delays or normal language development (Kim et al., 2014). Some children make speech and grammatical errors, such as incorrect articulation of consonants (Shriberg et al., 2001), misuse of personal pronouns (e.g., "she wants water" instead of "I want water"), or make errors in other syntactical rules (Kim et al., 2014). These deficits may be related to cognitive ability, too, however (Eigsti et al., 2011). Some children also exhibit prosody oddities, like flat affect or tone (Diehl & Paul, 2013; Lord & Paul, 1997), misplaced stress, slowed phrasing (Shriberg et al., 2001), and/or inappropriate volume and alternation between volumes (Shriberg, Paul, Black, & Van Santen, 2011). These speech oddities can also affect comprehension, as children with ASD may have trouble understanding others' intonations, prosody marks of questions, or emotion, or they may struggle to integrate knowledge and context with verbal stimuli (Diehl & Paul, 2013; Kim et al.,

2014). Again, there is heterogeneity in the domain of deficits and many of these deficits only apply to subsets of children with ASD.

As noted above, and contributing to heterogeneity in presentation, a subset of children with ASD do not develop spoken communication. Some children never develop spoken communication while others may have initially talked, and then ceased to do so. Cases of "regression," or lost skills, are commonly reported in the media, but, because studies are largely based on retrospective reports, more research is needed to examine the validity of these reports (Thurm, Powell, Neul, Wagner, & Zwaigenbaum, 2017). Importantly, many children who do not use spoken communication may be taught to communicate using sign language, pictures, or other methods of augmentative communication (Paul, 2009). For more information on teaching communication, see Chaps. 7 and 8.

1.1.2.3 Adaptive Functioning Deficits

In addition to the deficits that may disrupt social engagement, children with ASD may have motor delays and may be less likely to independently engage in daily living skills. The impairments in communication and social skills previously described likely contribute to adaptive skill deficits. Neurotypical children usually exhibit adaptive living skills that are aligned with their verbal or intellectual ability, but children with ASD may not (Klin et al., 2007). Children with ASD's adaptive functioning skills are often significantly below their measured cognitive ability (Kanne et al., 2011). The discrepancy between IQ and adaptive functioning is especially pronounced among individuals with high-functioning ASD, who often do not show improvements in adaptive living skills that are comparable to same-aged peers (Klin et al., 2007). These adaptive functioning deficits may manifest as an inability to independently dress, develop appropriate sleep hygiene, become toilet trained, or complete chores. Motor deficits often include difficulties with gross motor skills, like running or jumping, and fine motor abilities, like holding a pencil or tying shoes (Volkmar, 2013). Adaptive skills affect the everyday functioning of children with ASD across contexts including home, school, and the community.

In sum, children with ASD exhibit a wide array of difficulties in the two core domains that distinguish the diagnosis from others, but they also may demonstrate deficits in other areas, including cognition, language, emotion, and adaptive functioning. No two children with ASD have the same strengths, weaknesses, or presentations because even if they technically meet similar diagnostic criteria, the presentation and severity vary drastically. As our understanding of the presenting problems and the relation between deficits change, so too do the diagnostic criteria and diagnostic considerations. Many of the DSM-5 diagnostic criteria relate to the original case studies on ASD, but much of our understanding has and continues to change.

1.2 History

The current diagnostic criteria for ASD represent a historical development from the first case studies. ASD was first described in case studies by two independent researchers, Leo Kanner and Hans Asperger. In 1943, Austrian-American psychiatrist Leo Kanner met a 5-year-old child who took no interest in people around him, liked to spin around in circles, and threw tantrums when his typical schedule was interrupted (Kanner, 1943; Morrier, Hess, & Heflin, 2008). This case inspired Kanner to conduct 11 case studies, which he compiled into his groundbreaking paper, *Autistic Disturbances of Affective Contact* (1943). Kanner's paper was the first to differentiate "infantile autism" from "childhood schizophrenia," arguing this disorder was not "a departure from an initially present relationship" (p. 242). Rather, it was an "extreme autistic aloneness" (p. 242) in which the child does not respond to anything in the outside world. Kanner stated that the fundamental marker of autism was the "children's inability to relate themselves in the ordinary way to people and situations from the beginning of life" (Kanner, 1943 p. 242).

One year after Kanner's publication, Hans Asperger independently wrote about a

comparable, yet higher functioning disorder that he called "autistic psychopathy." Asperger similarly described children with social impairments and repetitive and restrictive interests and behaviors, but the children in his case studies had average language ability and above-average intelligence. Asperger was also the first to note abnormalities in nonverbal communication, describing behaviors such as a lack of eye contact and oddities in speech tone (Frith, 1991). Asperger also distinguished the children he observed from those with "childhood schizophrenia, noting that children with 'autistic psychopathy' did not have periods of normal development and their social impairments were stable, unlike children with schizophrenia" (Klin, 2011). While Asperger's case studies were eventually integrated with Kanner's in diagnostic systems, Asperger's work was largely lost until after World War II because he had published it in German in Austria during the war (Klin, 2011).

Kanner and Asperger both chose variants of the word "autism" and compared their findings to schizophrenia because that terminology and disorder were foci of the psychiatric literature at that time. In the early 1900s, schizophrenia included diagnostic criteria of egocentrism and social detachment that were called "autism" from the Greek root *autos,* or "self." Kanner and Asperger believed that their discoveries were separate from schizophrenia, but when "infantile autism" was entered into the World Health Organization's diagnostic system, the International Classification of Diseases (ICD), in 1967, it was considered a type of schizophrenia (Sasson, Pinkham, Carpenter, & Belger, 2010). Similarly, in the United States, the American Psychiatric Association's (APA) Diagnostic Statistical Manual (DSM) included "autistic" behaviors as a sign of childhood schizophrenia, but not a separate disorder (APA, 1968).

It was not until the 1970s that researchers distinguished autism from childhood schizophrenia. The distinction was made because autism and schizophrenia rarely occurred in the same families (Rutter, 1968) and had different developmental trajectories (Kolvin, 1971). These empirical findings led to a novel category of "Pervasive Developmental Disorders (PDD)" with a diagnosis of "autism" in the DSM-III in 1980 (Sasson et al., 2010). The criteria were later refined and expanded beyond Kanner's descriptions to include a required onset before the age of 3 of limited social responsiveness, language deficits, and/or "peculiarities," and "bizarre" environmental responses. The disorder was also distinguished from schizophrenia by requiring that individuals did not exhibit delusions, hallucinations, loose associations, or incoherence (APA, 1987).

The DSM-III diagnosis of autism did not capture all individuals who presented with similar characteristics, so PDD not otherwise specified (PDD-NOS) and Asperger's syndrome were added to subsequent versions of the DSM (Klin, 2011). A diagnosis of PDD-NOS was given when children did not meet full criteria for autism but still displayed impairing and distressing social, communication, and restrictive, repetitive behaviors (Ousley & Cermak, 2014; Volkmar, 2013). Asperger's syndrome was added to capture higher functioning cases of children who met the same criteria for social impairment and restricted, repetitive behaviors or interests as children with autism, but in the absence of language, cognitive, and adaptive behavior delays (APA, 1994). While the increase in diagnostic specificity and categories helped individuals receive the services and treatment they needed, it also led to a stark increase in the prevalence of diagnosed developmental disabilities (Zablotsky, Black, Maenner, Schieve, & Blumberg, 2015).

Research on the DSM-IV, PDD diagnoses did not support the separation of three diagnostic categories, however, which is why the DSM-5 integrated autism, Asperger's syndrome, and PDD-NOS into autism spectrum disorder (ASD; APA, 2013). The ASD diagnostic label and criteria were created to reflect research revealing that individuals with any of these diagnoses demonstrated behavioral variations of the same difficulties, rather than categorically different problems (APA, 2013). Furthermore, research supported that separation of the disorders did not result in reliable diagnoses across sites and did not predict the degree and severity of the disorder, prognosis, and treatment needs (Wiggins, Robins, Adamson,

Bakeman, & Henrich, 2012). Therefore, the diagnoses were combined but recognition of the variation and degree of severity was preserved by including ratings of severity that accompany the diagnosis. The severity rating system uses a three-point scale, with level one indicating lower levels of support and level three indicating "very substantial support" (American Psychiatric Association, 2013).

Of note, the ICD-10, which is used by American insurance companies, still includes social and communication as separate deficits and autism, Asperger's, and PDD-NOS as separate diagnoses because the ICD has not been updated since 1992. It is expected that the ICD-11 in 2018 will be similar to the DSM-5 (World Health Organization, 2017). And, even with different diagnostic systems, the presentation and prevalence of ASD are similar worldwide. However, the prevalence rates of ASD have fluctuated with variation in diagnostic criteria over time.

1.3 Prevalence

ASD is estimated to occur in 1% of the population internationally (American Psychiatric Association, 2013) and in about 1 in 59 people in the United States (Centers for Disease Control and Prevention (CDC), 2018). Boys are about four to five times more likely to be diagnosed with ASD than girls (APA, 2013), but girls are more likely to have a comorbid intellectual disability (ID) (Kirkovski, Enticott, & Fitzgerald, 2013). The male-to-female ratio difference has been consistent over the years, but the overall prevalence of the disorder has varied.

Estimates of ASD prevalence have increased substantially over the past several decades, from about 4.5 per 10,000 children in 1966 (Lotter, 1966) to about 146 in 10,000 children in 2006 (Centers for Disease Control and Prevention (CDC), 2015). The increase in prevalence rates is not thought to be due to an actual rise in the number of individuals diagnosed with the disorder; rather, it is hypothesized to be more due to modification of diagnostic criteria (King & Bearman, 2009) and an increase in knowledge and awareness of the disorder (Fombonne, 2005).

As previously described, the diagnostic criteria for ASD have changed over time, which affected both terminology and identification of individuals with ASD. As previously mentioned, the combination of autistic disorder, Asperger's syndrome, and PDD-NOS in the DSM-5 resulted in more individuals with an "autism" diagnosis (American Psychiatric Association, 2013). Additionally, diagnostic criteria for ASD, intellectual disability (ID), and other disorders were broadened, so more individuals met the criteria for ASD.

Changes to the ID diagnosis, formerly called mental retardation, may have also contributed to the rise in prevalence of ASD because the DSM used to specify that children could not have ASD and ID. Now, ID and ASD can be diagnosed together (APA, 2013), which directly increased the prevalence of ASD (King & Bearman, 2009). Some even hypothesize that individuals who currently meet the criteria for ASD were previously mislabeled as having ID, as evidenced by the fact that the prevalence of ID alone decreased while the prevalence of ASD increased. While support for the hypothesis that mislabeling contributed to prevalence changes is mixed due to the lack of causal data, it is clear that changes to diagnostic criteria for ASD and ID led to more ASD diagnoses (King & Bearman, 2009).

Another result and potential contribution to the increased prevalence was improved access to diagnostic and intervention services. These improvements were in part due to the already increasing prevalence, because public health initiatives, policy, and law were launched to increase knowledge, awareness, and acceptance of the disorder. Organizations such as the Centers for Disease Control (CDC) and Autism Speaks created initiatives to disseminate information on signs, treatments, and resources for the disorder. Other groups, such as the National Research Council (2001) and the American Academy of Pediatrics (Johnson & Myers, 2007), recommended screening tools to facilitate diagnostic evaluations at a younger age. ASD was also included as a category in special education law in

1991, which led to diagnoses and accommodations in school settings. School-based diagnoses and referrals are particularly important for families without regular access to healthcare (Gurney et al., 2003). Between the public health, policy, and legal initiatives, information on ASD is more readily available, which helps parents, doctors, school personnel, and others identify early signs and seek appropriate referrals for diagnoses of ASD and comorbid conditions (Wing & Potter, 2002).

1.4 Comorbidities

1.4.1 Psychiatric Conditions

Comorbid conditions affect the presentation, diagnosis, and treatment of ASD, so they are an important consideration when assessing the presentation and needs of children with ASD and their families. Current estimates suggest that approximately 70% of individuals with ASD have at least one comorbid diagnosis and about 40% of those individuals have more than two comorbid diagnoses (American Psychiatric Association, 2013). However, these statistics should be interpreted with caution as there are no current prevalence studies on comorbidities using DSM-5 criteria and diagnostic boundaries are not well understood. ASD behavioral indicators overlap with many psychiatric and medical signs and comorbid diagnoses have low reliability across practitioners. Therefore, it can be difficult to determine when an individual's behaviors indicate distinct conditions and when they are related to the core deficits of one disorder (Gurney, McPheeters, & Davis, 2006). For example, children with ASD may avoid or isolate themselves while in social situations. This presentation may be consistent with a peer's presentation of social anxiety, while a child with ASD's social impairment must be taken into consideration when determining if a differential diagnosis is appropriate.

Social anxiety and other anxiety disorders are some of the most commonly comorbid diagnoses with ASD, especially among school-aged children and adolescents. Estimates of anxiety among children with ASD range from 42 to 79% of children (Kent & Simonoff, 2017). The most common subtypes of anxiety in children with ASD are specific phobias, which are estimated to occur in about 31 to 67% of children with ASD, and social anxiety, which likely occurs in about 4 to 29% of children with ASD (Kent & Simonoff, 2017). Reports on the prevalence and presentation of anxiety with ASD are inconsistent, in part because anxiety disorders are difficult to diagnose in children in general and in children with ASD, specifically. Because children with ASD demonstrate deficits in emotional regulation and social-communication impairment is a core deficit in ASD, it can be difficult to differentiate when a comorbid anxiety diagnosis is warranted. Additionally, anxiety is also difficult to be identified by caregivers because it is more of an internal experience (Ryan, Kaskas, & Davis, 2017). As a result, diagnosis and research on anxiety in children with ASD are difficult and findings are inconsistent. But, when looking at reviews across studies, it seems most likely that individuals across the ASD spectrum are at risk for comorbid anxiety (e.g., Kerns & Kendall, 2012).

The type and severity of anxious thoughts and behaviors seem to be fairly similar between children with and without ASD, but there are a few differences. For example, the most common phobias among neurotypical children involve animals or death and injury. And, while some children with ASD share these fears, children with ASD are more likely to report phobias and fears about things such as mechanical objects (e.g., toilets) or the weather (e.g., refusal to go outside on cloudy days; Mayes et al., 2013). Another example is that some children with ASD and social anxiety report concerns about social evaluation that are similar to those of neurotypical children, but others are avoidant and meet the criteria for social anxiety without social evaluation concerns (Kerns & Kendall, 2012), which may be in part due to children's (with ASD) difficulty taking another person's perspective and interpreting their behaviors.

More obvious comorbid psychological conditions that co-occur with ASD at similar rates as anxiety are disruptive, externalizing behaviors, such as aggression, self-injury, noncompliance, or destruction of property, as will be covered in more detail in Chaps. 10 and 11. Disruptive externalizing behaviors and disorders, which are sometimes diagnosed as oppositional defiance disorder (ODD) or conduct disorder (CD), occur in approximately one-fourth (Hill et al., 2014) to one-half of children with ASD (Mazurek, Kanne, & Wodka, 2013). These disruptive behaviors pose physical and emotional risks for the child and others. Some children harm themselves through head-banging, biting their own skin, or other self-injurious behaviors. Some engage in aggressive hitting, kicking, or biting of others. Some are disruptive by throwing objects, breaking items, or throwing tantrums, while others may be noncompliant with directions or rules. Children may also exhibit several of these behavioral topographies.

Disruptive behaviors are especially important areas of intervention for children with ASD due to the risk of physical harm, destruction, and caregiver stress and burnout (Woodman, Mawdsley, & Hauser-Cram, 2015). Problem behaviors often do not decrease naturally with age for children with ID and ASD (Matson & Shoemaker, 2009), though such children are at higher risk for these co-occurring problem behaviors (e.g., Hill et al., 2014; McTiernan, Leader, Healy, & Mannion, 2011). Some also report that children with less verbal communication are at higher risk for aggression (e.g., Farmer et al., 2015), but that relation is not consistently found (Hill et al., 2014; Kanne and Mazurek, 2011), and may be accounted for by other characteristics (e.g., Mazurek et al., 2013). In general, children who are more likely to need long-term care and support are at higher risk for disruptive behaviors that interrupt daily life and learning opportunities, and can lead to further isolation for the child and family (Horner et al., 2002), which is why the behavioral interventions that will be covered in Sects. 1.1 and 1.2 of this book are a high priority.

Another common comorbidity is attention-deficit hyperactivity disorder (ADHD), which co-occurs with ASD in about 40–80% of cases (Reiersen & Todd, 2011). ADHD could not previously be diagnosed with ASD due to the overlap in executive functioning deficits that are common among children with ASD (APA, 2000). However, changes to the DSM-5 now provide the flexibility of the comorbid ASD and ADHD diagnoses. It is difficult to differentiate executive functioning deficits due to ASD and those due to ADHD, so a child is diagnosed with ADHD if they meet the criteria for inattention (e.g., trouble attending to or remembering lessons or instructions, difficulty with organization or planning), hyperactivity (e.g., frequent fidgeting or climbing), and/or impulsivity (e.g., interrupting conversations, trouble waiting turns) that are not developmentally appropriate (APA, 2013). For example, even though many children with ASD like to adhere to schedules, a child with ADHD and ASD may exhibit more trouble independently completing the schedule. The child may become distracted going between classes if they are unable to inhibit an impulse to join a more appealing activity. He or she may lose a schedule and forget what came next. Or, the child may have trouble completing activities because it can be hard to plan and execute multiple steps to reach a goal. Again, while ADHD and ASD deficits overlap, a comorbid diagnosis is often made to indicate that additional supports are needed.

As with ADHD, differential diagnosis with all psychiatric conditions is complicated due to limited understanding of the etiology, relation between disorders, and imprecise measurement. As a result, diagnosticians and clinicians should recognize the limitations and benefits to making comorbid psychiatric diagnoses. The diagnoses are likely to have low reliability and may not represent two separate disorders. The diagnostic criteria and practices are also continually improving as the research does, so it is important to stay current on changes in the field. Benefits include that identification of impairing and distressing psychological problems can help the child and family get needed services.

1.4.2 Developmental and Physical Conditions

Children with ASD may also require medical services to address commonly comorbid genetic and physical conditions, like genetic syndromes, motor deficits, feeding disorders, and medical diagnoses (e.g., gastrointestinal problems, epilepsy, sleep problems; APA, 2013). As with psychological disorders, the overlap between genetic and physical conditions with ASD is not well understood, but the medical diagnoses are more clearly differentiated from ASD.

Genetic disorders can be reliably diagnosed because there are clear biological markers. There is also a possible common etiology between the genetic disorder and ASD as children with ASD are at a higher risk for hundreds of genetic syndromes (Schaefer & Mendelsohn, 2008). Some of the most commonly known co-occurring genetic disorders are fragile X, tuberous sclerosis, Rett syndrome, Prader-Willi syndrome, Angelman syndrome, and Down syndrome. Many of these disorders also have co-occurring IDs, so genetics may account for the link between ASD and ID in some of these cases (Zafeiriou, Ververi, Dafoulis, Kalyva, & Vargiami, 2013). In addition to genetic syndromes, children with ASD are also at increased risk for other physical difficulties, such as motor impairments. Motor impairments in ASD include the ability to integrate information to plan and execute motor tasks (Gowen & Hamilton, 2013), low muscle tone, deficits in the execution of skilled movements (e.g., drooling, inability to use scissors), and intermittent toe-walking (Ming, Brimacombe, & Wagner, 2007). Motor deficits affect the child's adaptive living skills, like independently completing tasks, writing, or engaging in normal, childhood recreational activities.

In addition to genetic and motor impairments, children with ASD are also at an increased risk for feeding and gastrointestinal problems. Children with ASD have a fivefold increase in the odds of developing a feeding problem compared with peers (Sharp, Berry, McElhanon, & Jaquess, 2013). Feeding difficulty in children with ASD most commonly presents as food selectivity, involving strong preferences for starches and snack foods, limited consumption of fruits and vegetables, selectivity based on the texture of food, and mealtime difficulties like disruptive mealtime behavior. The feeding problems may be related to gastrointestinal problems, such as constipation and related discomfort (McElhanon, McCracken, Karpen, & Sharp, 2014), behavioral rigidity and sensory sensitivity (Ahearn, Castine, Nault, & Green, 2001; Provost, Crowe, Osbourn, McClain, & Skipper, 2010), or a learned pattern of escape and/or attention when refusing food during mealtimes (Piazza, 2008; Piazza et al., 2003).

Finally, other common medical problems in children with ASD include epilepsy and sleep problems. Epilepsy is estimated to occur in 5–38% of children with ASD. Seizures also require close parental and medical monitoring to ensure safety for the child. And, because seizures cause neurological deficits, they can also contribute to behavioral problems, like increased impulsivity (Levisohn, 2007). Maladaptive behaviors, like impulsivity, aggression, decreased adaptive functioning, and executive functioning deficits, are also exacerbated by commonly co-occurring sleep problems for children with ASD. Sleep problems that are elevated in children with ASD include difficulty falling asleep, less time spent sleeping due to frequent, long waking in the middle of the night and/or waking early, more frequent night terrors, and more anxiety about sleep (Souders et al., 2009). An estimated 50–80% of individuals with ASD have sleep problems, which increase the risk of aggression two- to threefold (Mazurek et al., 2013). As with the other comorbid conditions, child sleep problems also disrupt the families' sleep cycle and routines.

Mental and physical health comorbidities are important for clinicians to consider due to the impact they have on the child and family. Mental and physical health conditions contribute to the heterogeneity in presentation of ASD and they often exacerbate or contribute to the development of other presenting concerns. Therefore, identification of areas of impairment and/or distress is an important consideration for treatment. To

improve identification of comorbid diagnoses, it is important to understand the underlying etiology of ASD that can lead to a wealth of knowledge and potentially contribute to a better understanding of related and/or comorbid behavioral indicators. To date, there are some hypotheses about the etiology that can be helpful in predicting the risk of ASD and other conditions.

1.5 Risk Factors and Etiology

While the etiology of ASD is not well understood, it is believed to be strongly genetically determined. But environmental factors also contribute directly and through their interaction with genetics and biology (Bailey et al., 1995; Lichtenstein, Carlstrom, Rastam, Gillberg, & Anckarsater, 2010). Risk factors have been more consistently identified than causes, so genetic and environmental risk factors with the most support are reviewed below.

1.5.1 Genetic and Biological Findings

Evidence for the genetic basis of ASD comes from studies documenting a higher risk for the disorder among family members of someone with ASD. Studies that demonstrated higher concordance rates for ASD in monozygotic twins, compared to dizygotic twins, estimated that genetic effects account for about 37 (Hallmayer et al., 2011) to about 90% of the proportion of variance in ASD (Geschwind, 2011). Siblings of a child with ASD are also 13–22 times more likely than the general population to also be diagnosed with ASD (Lauritsen, Pedersen, & Mortensen, 2005). In addition to higher rates of the disorder among close relatives, there are also higher rates of genetically and qualitatively related behaviors, labeled the "Broader Autism Phenotype" (Losh et al., 2009). The Broader Autism Phenotype consists of milder, subthreshold abnormalities in restrictive, repetitive behaviors and social-communication functioning, which manifest in characteristics that have been termed aloofness, rigidity, and pragmatic language (Seidman, Yirmiya, Milshtein, Ebstein, & Levi, 2012). About 12–30% of family members of individuals with ASD are estimated to be rated as high in the Broader Autism Phenotype (Cruz et al., 2013). But, some individuals with ASD are the only affected family members and the genetics and transmission patterns seem to be different dependent on whether one or many family members have the disorder (Sebat et al., 2007). In families in which multiple family members are affected, common variations of genes each has small additive impacts on the development of ASD (Constantino, Zhang, Frazier, Abbacchi, & Law, 2010; Gaugler et al., 2014). In families in which only one child has ASD, however, rare mutations to DNA sequence likely play bigger roles in risk for the disorder (Sebat et al., 2007).

The genetic etiology that contributes to ASD is as variable as the presentation. Thousands of candidate genes (segments of DNA that are risk factors), DNA sequence abnormalities, chromosomal rearrangements, and single-gene mutations have been identified as possible contributors to the etiology of ASD; however, each abnormality only accounts for approximately 1% of cases (Betancur, 2011; Marshall et al., 2008; Weiss, 2009). The identified susceptibility genes only account for about 10–20% of all ASD cases (Geschwind, 2011). Because the list of genetic findings is long and research is ongoing, a full list of genetic findings for ASD will not be included here. An up-to-date list of genetic findings and supporting research can be found at online databases by SFARI (http://www.gene.safari.org) or AutDB (http://www.mindspec.org). The current section focuses on remaining questions, overarching conclusions, and some of the genetic findings with the strongest, most consistent support.

The genetic abnormalities that are associated with ASD are believed to affect brain, physiological, and metabolic functioning, but the mechanism of contribution to the disorder and function of the mutations are unknown (Weiss, 2009). The identified and replicated genes associated with ASD are known contributors to neural

connectivity (Persico & Napolioni, 2013) and physiological and metabolic systems, like immune and inflammation systems (Rossignol & Frye, 2012). The abnormalities are expected to combine and interact with each other and the environment to disrupt neural network (Geschwind & Levitt, 2007) and to cause damage to brain and cell tissues (Rossignol & Frye, 2012; Streit, Mrak, & Griffin, 2004). But, it remains unclear if the identified genes cause ASD, how large the effect has, if the genetic abnormalities are deterministic, or if the findings are due to chance (Geschwind, 2011). Therefore, more research on the role and function of the genetic findings is needed.

Further complicating genetic findings is the overlap in genetics between ASD and other psychological disorders. Genes that have been associated with ASD were also associated with schizophrenia (Kim et al., 2008), mental retardation, cardiac abnormalities, and abnormal head sizes (e.g., Brunetti-Pierri et al., 2008). Genetics also contribute to concordance in comorbidity for monozygotic twins. The overlap between disorders suggests a common genetic etiology underlying multiple disorders (Lichtenstein et al., 2010).

In sum, genetic research has identified some important genes that may contribute to ASD, but the impact of these mutations and gene variants is not well understood and requires further evaluation. The common variants of genes that are expected to play an important role have been challenging research and replicate (Geschwind, 2011). Thus, research on the genetic etiology of ASD is ongoing but guided by the findings that the genetic etiology is as heterogeneous as the presentation of the disorder and gene-environment interactions all need to be further explored.

1.5.2 Environmental Risk Factors

There are many possible environmental risk factors for ASD that have been identified, but the mechanisms of action are not yet well understood and the correlations with ASD are often weak. These environmental factors that have been documented to increase the risk of ASD can occur at conception, during pregnancy, and during birth. Many of these risk factors co-occur, so the interaction between them may also be important (Mandy & Lai, 2016).

Beginning at the start of the child's life, risk factors at conception and during pregnancy that affect fetal development include parental age, maternal mental and physical health, and maternal exposure to environmental and psychotropic substances. Risk factors during birth include birthing complications and physical health abnormalities (Mandy & Lai, 2016).

At conception, both parents' ages have been observed to have cross-sex effects on the risk of ASD. The risk of ASD increases twofold for every 10-year increase in paternal age, especially if the child is a female (Sandin et al., 2012). This means that children born to a father between the age of 30 and 39 are twice as likely to have ASD than if the father was between the age of 20 and 29. Children conceived by men over the age of 40 are 5.75 times more likely to develop ASD (Reichenberg et al., 2006). This effect is possibly due to a mutation in spermatozoa. Spermatozoon cells replicate every 16 days throughout the life span. Each cell division increases the possibility of a replication error and gene mutation, so a longer life span might mean more replication errors (Mandy & Lai, 2016). Similar to fathers, mothers over the age of 35 are twice as likely to have a child with ASD, especially if the child is male. This effect may be because older mothers have a longer history of exposure to other environmental risk factors that contribute to developmental delays (Sandin et al., 2012). In addition, a meta-analysis found increased ASD risk in first-born children compared to those born third or later (Gardener, Spiegelman, & Buka, 2011).

Other maternal characteristics that affect the risk of ASD include maternal mental and physical health. Maternal, but not paternal, history of psychopathology is associated with a twofold increased risk for ASD (Lauritsen et al., 2005). Intake of the psychotropic medications used to treat psychopathology while pregnant also increases the risk of offspring with ASD. For example, maternal intake of antiepileptic drugs

or valproates during pregnancy increases the risk of having an offspring with a neurodevelopmental disorder, such as ASD. Valproates treat epilepsy, bipolar disorder, and migraines (Bromley et al., 2013). Selective serotonin reuptake inhibitors (SSRIs), which are medications for depressive disorders, also have a link to high-functioning ASD (Mandy & Lai, 2016). It is unclear if both medications increase the risk of ASD or if there is a common connection between maternal psychopathology and child ASD. The cause fueling the relationship between these medications and a diagnosis of neurodevelopmental disorders is still unclear, as is the effect on the level of functioning of the offspring (Bromley et al., 2013).

In addition to maternal mental health, maternal physical health affects the likelihood of offspring ASD. Maternal obesity (Li et al., 2016) and diabetes (Xu, Jing, Bowers, Liu, & Bao, 2014) during pregnancy also increase the risk for many psychosocial and developmental problems, including ASD. This connection could be linked to an increase in glucose levels in diabetic and prediabetic women that adversely affects fetal development. Prolonged fetal exposure to glucose causes the fetus to increase oxygen consumption and metabolism, which may result in a fetal iron deficiency and subsequent neurodevelopmental abnormalities. Fetal oxygen and neurodevelopment can also be affected by birthing events and traumas (Tanne, 2012).

Risk factors for ASD associated with birth include hemorrhaging, complications related to the umbilical cord, and multiple births. Additionally, children who are born at a low birth weight in general or for the baby's gestational age have abnormal breathing, birth defects, trouble feeding, anemia, hemolytic disease, and hyperbilirubinemia and are also at increased risk for ASD (Gardener et al., 2011). Again, these environmental factors are probabilistic, not deterministic, so not all children who experience these conditions will develop ASD. But, there are some known environmental factors that were originally called causal and have since been reported to be unrelated to the development of ASD. Because they have received so much attention in mainstream media, they will be described below.

1.5.3 Myths About Causes of ASD

Although there is still more to learn about the etiology and risk factors contributing to the development of ASD, we can dispel some previously believed myths, which will also be explored in more detail in Chap. 6. One of the first myths was that "refrigerator mothers," or emotionally unavailable mothers, caused ASD (Allen, DeMyer, Norton, Pontius, & Yang, 1971). This myth was formed by Dr. Leo Kanner, the first doctor to describe ASD in 1948. Kanner noted that the children he observed with his newly defined disorder seemed to have mothers who lacked warmth and attachment. He then concluded that these "refrigerator mothers" caused the social, communication, and behavioral abnormalities. What Kanner did not consider, however, was that his sample size was small; many "refrigerator mothers" did not have children with ASD, and the lack of warmth and attachment may have been a result of the ASD-related behaviors, instead of the cause (Laidler, 2004). Empirical research does not support Kanner's theory, but mother-blaming lasted for decades which as a result harmed countless families of children with ASD.

Another commonly cited myth is that measles, mumps, and rubella (MMR) vaccinations cause ASD (Wakefield et al., 1998). The myth arose when Dr. Andrew Wakefield published research linking the MMR vaccine to ASD and claimed the vaccine did not undergo proper safety testing (Wakefield et al., 1998; Wakefield & Montgomery, 2000). His articles caused a public health crisis in many countries because an international measles outbreak occurred, after parents refused to vaccinate their children (Flaherty, 2011). Dr. Wakefield came under investigation a few years later, when his findings failed to replicate and the rates of ASD diagnoses continued to increase in areas that did not administer vaccines (Honda, Shimizu, & Rutter, 2005). The investigation demonstrated that Dr. Wakefield falsified his data to personally benefit from the findings. And, even though he was found guilty of ethical, medical, and scientific misconduct, the article was retracted, and empirical research disproved his claims, some parents still believe that MMR vac-

cines cause ASD and refuse to vaccinate their children (Flaherty, 2011).

Finally, a more recent proposal that resulted in public action without empirical substantiation states that gluten and casein contribute to the development and severity of ASD. This theory is based on findings that children with ASD have higher levels of gluten and casein peptides in their urine. Normally, gluten and casein foods should be broken down into amino acids. If those foods are not properly broken down, then larger chains of amino acids, called peptides, remain. Because children with ASD seem to have higher levels of these peptides, some theorized that children with ASD lack the enzymes needed to break down gluten and casein foods. It was further proposed that the peptides build up, enter the bloodstream, cross the blood-brain barrier, attach to opioid neuroreceptors, and change the brain in ways that cause ASD (Reichelt, Knivsberg, Lind, & Nodland, 1991). This is a relatively new theory that received some support, but scientists are still skeptical because gluten and casein interventions have not been heavily, nor rigorously, researched. The few studies that exist did not include control groups, relied on small samples or anecdotal evidence, and had many risks of biases, such as confirmation bias or placebo effect. Despite the lack of evidence supporting this notion, some parents have been prescribing to these restrictive diets, which may negatively impact the child's health by depriving them of nutrients and may cause added stress to the family (Marí-Bauset, Zazpe, Mari-Sanchis, Llopis-González, & Morales-Suárez-Varela, 2014; Millward, Ferriter, Calver, & Connell-Jones, 2009). The gluten and casein intervention requires more rigorous study because, like all interventions, it can be time and resource intensive for families of children with ASD. Family factors are especially important to consider because ASD and related accommodations have a particularly high impact on families.

1.6 Impact on the Family

Because ASD is a lifelong disability, individuals on the spectrum need more intensive care for longer periods of time than individuals without ASD. The types of care include educational, occupational, medical, psychotherapeutic, and familial care to accommodate behavioral, social, and emotional difficulties (Symon, 2001). A review of longitudinal studies on adaptive living skills for individuals with ASD reported that daily living skills often improved, but social skills and communication did not. Fortunately, many individuals with ASD are able to achieve some degree of independence, to hold a job, and to make friends, but about 50% of adults with ASD remain dependent on caregivers and special services for living arrangements and employment (Magiati, Tay, & Howlin, 2014).

The cost for caregivers and on society is substantial, with estimates reaching 17,000 dollars more per year for a child with ASD than without (Lavelle et al., 2014) and about 3.2 million dollars over the life span (Ganz, 2007). The largest expense for children with ASD is school services as children with ASD are approximately nine times more likely to need an individualized education program (IEP) and about eight times more likely to need some form of special education compared to children without ASD (Lavelle et al., 2014). Another substantial expense for children with ASD is medical care, particularly between the ages of 3 and 7. Direct medical care for the first 5 years can cost on average $35,000 per year, not including the cost of alternative treatments or adverse outcomes due to failed treatments (Ganz, 2007).

Lost productivity for parents is a major concern because it can lead to decreased income and increased stress level. Mothers of children with ASD report spending more time and money to obtain care for their children than parents of children with other special healthcare needs (Kogan et al., 2008). One study found that mothers of children with ASD who were eligible for higher paying jobs (e.g., had the highest educations and were older) earned approximately 35% less than mothers of children with other health limitations and about 56% less than mothers of children with no health limitations. Fathers' work schedules were less likely to be impacted, but families of a child with ASD still had a lower household income than families with neurotypical children (Cidav, Marcus, & Mandell, 2012). And, unfortu-

nately, despite the extra time and effort, parents of children with ASD were less likely to report that their healthcare needs were met or to feel satisfied with services than mothers of children with other special healthcare needs (Kogan et al., 2008).

The dissatisfaction with care, increased need for care, child's behaviors, and contextual factors contribute to higher levels of stress for parents of children with ASD than for parents of neurotypical children (Duarte, Bordin, Yazigi, & Mooney, 2005), parents of children with other psychopathological disorders (Lee, Harrington, Louie, & Newschaffer, 2008), and parents of children with other developmental delays (Estes et al., 2009). In addition to the extra money, time, and energy needed to care for and provide services for the child, the child's comorbid diagnoses (Bebko, Konstantareas, & Springer, 1987; Plant & Sanders, 2007) and adaptive functioning deficits have also been reported to contribute to parental stress (Hall & Graff, 2011). Parental stress also may further exacerbate difficult behaviors associated with ASD, resulting in a bidirectional, perpetuating cycle (Karst & Van Hecke, 2012).

Other outcomes of and contributors to parent stress are physical, emotional, and psychological problems for the parent, which results in more stressors to the family (Brobst, Clopton, & Hendrick, 2008). While the direction of effects is unclear, parents of children with ASD, especially mothers, have higher levels of depression and anxiety (Olsson & Hwang, 2001; Hastings, 2003), and physical health problems, even compared to parents of children with other disabilities (Mugno, Ruta, D'Arrigo, & Mazzone, 2007). These psychological and physical health conditions can make it harder to care for the child and can also have a negative effect on the family.

Fortunately, stress associated with parenting a child with ASD does not generally affect parent's perceived closeness of the relationship with the child, but it may affect marital satisfaction and status (Hoffman, Sweeney, Hodge, Lopez-Wagner, & Looney, 2009). Parents of children with ASD are twice as likely to get divorced as parents of neurotypical children (Hartley et al., 2010). Parents of children with ASD who stay married report lower levels of marital satisfaction compared to parents of children with other developmental delays or no disabilities (Santamaria, Cuzzocrea, Gugliandolo, & Larcan, 2012). Further, divorce and marital discord often result in lower levels of financial and social support, which are protective factors against stress and psychopathology (Bromley, Hare, Davison, & Emerson, 2004).

Some research has identified protective factors that minimize stress for parents of children with ASD. For example, parental use of coping strategies that focus on seeking social support, problem-solving, focusing on the positive, expressing emotions in a constructive way, and compromising has been shown to improve mood and minimize stress (Pottie & Ingram, 2008). Additionally, parents who are lower in the Broader Autism experience lower levels of stress (Ingersoll & Hambrick, 2011). Thus, it can be important to increase parent's utilization of and/or access to social support and coping strategies to decrease stress.

The impact of ASD on siblings has received less research attention and extant studies differ somewhat in outcomes. For example, in some studies siblings report overall positive relationships with their sibling with ASD and positive self-concepts (e.g., Macks & Reeve, 2007) and are not at increased risk for adjustment difficulties (e.g., Kaminsky & Dewey, 2002), yet other studies suggest that siblings are at risk for internalizing and externalizing problems, and report negative relationships with their sibling with ASD (e.g., Meadan, Stoner, & Angell, 2010; Ross & Cuskelly, 2006), and increased risk of adjustment difficulties (Fisman, Wolf, Ellison, & Freeman, 2000). Some of the variability in findings may be due to methodological differences between studies and small samples, but some may be due to differences depending on the severity of the child with ASD's presentation, the sibling's characteristics, and family functioning (Meadan et al., 2010). Therefore, further research on the family system and impact on siblings is needed to understand factors that influence family functioning and sibling outcome. This

research should lead to interventions to help families remain intact, support one another, and access needed resources.

1.7 Conclusions

The criteria and our understanding of the etiology of ASD have changed over time, but with a wealth of past and ongoing research there are promising implications for future discoveries related to ASD. Improved understanding of and awareness for the disorder have resulted in a higher number of children diagnosed with ASD with 1 in every 59 children diagnosed with ASD in the United States. Boys are more likely to receive the diagnosis compared to girls and there are substantial variations in the presentation of the disorder across the sexes and within ASD more broadly. Children with ASD do not only face the abnormalities associated with their ASD diagnosis, but they are at an increased risk for other mental health and medical diagnoses as well. The etiology of the disorder may also encompass genetic, mitochondrial, or other medical diagnoses that further affect their presentation and affect the individual and his or her family. Families of children with ASD display higher stress, lower incomes, and many related psychosocial stressors associated with caring for a child with special needs. More research is needed to continue to learn about the heterogeneous diagnosis of ASD, but the recent influx of research on this population has led to many new discoveries and new pathways for investigation.

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