

Foundations of Autism Spectrum Disorders: An Online Course

Session 1

Understanding Pervasive Developmental Disorders and Autism Spectrum Disorders

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Session 1, Lesson A

Historical Background and DSM IV TR Classification of Pervasive Developmental Disorders

Upon completion of Session 1, Lesson A, learners will:

1. identify early theorists in the field of autism.
2. list the 5 diagnoses described in the DSM-IV-TR that comprise the category of pervasive developmental disorder (PDD).
3. describe the primary features required for a diagnosis of autistic disorder.
4. identify the distinctions between autistic disorder and Asperger's disorder.
5. list the PDD disorder that primarily occurs in girls and is caused by a genetic mutation.
6. identify the rare PDD diagnosis that is characterized by early normal development followed by regression that occurs after 2 years of age.
7. describe the criteria for a diagnosis of PDD-NOS.

Historical Background

Autism was first described in 1943 by Leo Kanner, a child psychiatrist at Johns Hopkins University in Baltimore, Maryland. He wrote about eleven children who were similar to each other but whose pattern of difficulties were quite different from children diagnosed with other conditions. His rich description and characterization of these children and of their disability have provided a firm foundation for subsequent work in the field. Because Dr. Kanner considered social withdrawal to be the primary feature of the disability, he used the term "autism," from the Greek word *autos*, meaning self, and called the disorder "early infantile autism". He observed that the children became upset when other changes to routines occurred (i.e., insistence on sameness), and often engaged in repetitive behavior, such as hand-flapping. Dr. Kanner (1943, 1949) also pointed out that the children had impairments in their language, including echolalia (i.e., repeating back what they have heard), literalness, and pronominal reversal (i.e., using "you" instead of "I" to refer to self). Although our understanding of autism has broadened, the initial triad of symptoms described by Leo Kanner more than 60 years ago still

represents the core symptoms that define autism: social isolation, repetitive behaviors and restricted interests, and communication and language impairments.

Some of Kanner's original writings can be found on the following Web site:
http://www.neurodiversity.com/library_kanner_1943.html.

Dr. Lorna Wing, a British researcher whose work in the field of autism has been influential, published a paper (Wing, 1981) to draw attention to the work of Hans Asperger. Dr. Asperger, an Austrian pediatrician, had published a paper in 1944 describing a group of individuals whose characteristics included little empathy for others, one-sided conversations, and intense interests. He used the term *autistic psychopathy* to describe the disorder. His papers, written in German and published during World War II, had gone unnoticed by most of the English-speaking world. Once published in English, Asperger's work (1944, translated by Firth, 1991) contributed to the recognition in the 1980s and 1990s that there were children and adults with a higher functioning form of autism than had typically been diagnosed. The diagnosis of Asperger's syndrome is now used to refer to a subgroup of individuals on the autism spectrum who have average or above-average intelligence and good verbal skills, but who may have difficulties with social interactions and/or restrictive, repetitive behaviors.

Some of Asperger's original writings can be found by linking to the following Web site:
<http://www.mugsy.org/wing2.htm>

Pervasive Developmental Disorders (PDD)

In the United States, the *Diagnostic and Statistical Manual of Mental Disorders*, or DSM, of the American Psychiatric Association provides the criteria used as the basis for the diagnosis of mental health and developmental disorders. The DSM is revised periodically, and the current edition, published in 2000, is the DSM-IV-TR. This version of the DSM is currently under review. The new version, DSM-5, is projected to be published in May 2013. Outside of the United States, the *International Classification of Diseases*, 10th edition (ICD-10) is widely used. The criteria for autism used in the DSM-IV-TR and the ICD-10 are similar though not identical (American Psychiatric Association, 2000; World Health Organization, 1992). This discussion is based on criteria set forth by the DSM-IV-TR. Please note that the forthcoming DSM-5 is expected to bring some changes to the categories and definitions discussed below.

The DSM is divided into many categories, including the major mental disorders and developmental and learning disorders. The autism spectrum disorders are developmental disorders and are classified under a heading called "Pervasive Developmental Disorders." The term *pervasive developmental disorders* (PDD) is considered by many to be synonymous with autism spectrum disorders, however, the DSM-IV-TR category includes five diagnoses—the three disorders referred to as autism spectrum disorders plus Rett's disorder and childhood disintegrative disorder. These

five disorders will be discussed in this section. The disorders included in this category all involve developmental differences across many areas of functioning. Significant impairment in social interaction is a core feature of each. For the criteria of each disorder go to: <http://www.cdc.gov/ncbddd/autism/hcp-dsm.html>

The five diagnoses in the pervasive developmental disorders category are:

- autistic disorder,
- Asperger's disorder,
- Rett's disorder,
- childhood disintegrative disorder, and
- pervasive developmental disorder not otherwise specified (PDD-NOS).

Autistic Disorder

Autistic disorder is the most common of the pervasive developmental disorders. The symptoms of autism often appear early in life and, by definition, occur prior to 3 years of age. Some parents report that differences were apparent in infancy (Landa, 2007; Werner, Dawson, Osterling, & Dinno, 2000). For other children, developmental differences may not have been noticeable until 18 to 36 months of age.

Individuals who have autism display difficulties in three areas: social relationships, communication, and restrictive or repetitive behaviors and interests. In each of these three areas, the specific difficulties that a person with autism might have can vary considerably. The DSM provides a list of four possible indicators of qualitative impairment in social interaction, four indicators of qualitative impairments in communication, and four indicators of restricted repetitive and stereotyped patterns of behavior, interests, and activities. In order to qualify for a diagnosis of autism, a person needs to have at least two of the social indicators, one of the communication indicators, and one of the restricted and repetitive interests or behaviors. As these criteria suggest, there is tremendous variability in the specific sets of impairments seen among individuals with autism. Some discussion and further description of these impairments and characteristics will be provided in a later section. The text box provides the specific criteria used in the DSM-IV-TR for each of these areas.

Asperger's Disorder

The diagnostic criteria for Asperger's disorder, as defined by the DSM-IV-TR, are quite similar to the criteria for autistic disorder. The specific criteria for social interaction difficulties and for restricted, repetitive and stereotyped behavior are exactly the same. Differences appear in the area of communication and in the requirements for cognitive and adaptive skills; in addition, onset does not have to have been apparent before the age of 3 years. Thus, the criteria for Asperger's disorder require that the child's early language development was not significantly delayed, that cognitive development is not delayed (i.e., that cognitive skills are in the average or above average range), and that adaptive behavior in all areas other than social interaction is not delayed. The criteria for the diagnosis of Asperger's disorder specifically exclude persons for whom the criteria

are met for another specific pervasive developmental disorder, including autistic disorder, or schizophrenia.

Diagnostic Criteria for Autistic Disorder

- A. A total of six (or more) items from (1), (2), and (3), with at least two from (1), and one each from (2) and (3):
1. qualitative impairment in social interaction, as manifested by at least two of the following:
 - a. marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
 - b. failure to develop peer relationships appropriate to developmental level
 - c. a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest)
 - d. lack of social or emotional reciprocity
 2. qualitative impairments in communication as manifested by at least one of the following:
 - a. delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
 - b. in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
 - c. stereotyped and repetitive use of language or idiosyncratic language
 - d. lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level
 3. restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:
 - a. encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
 - b. apparently inflexible adherence to specific, nonfunctional routines or rituals
 - c. stereotyped and repetitive motor manners (e.g., hand or finger flapping or twisting, or complex whole-body movements)
 - d. persistent preoccupation with parts of objects
- B. 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.
- C. The disturbance is not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder.

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Diagnostic Criteria for Asperger's Disorder

- A. Qualitative impairment in social interaction, as manifested by at least two of the following:
 - 1. marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
 - 2. failure to develop peer relationships appropriate to developmental level
 - 3. a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by a lack of showing, bringing, or pointing out objects of interest to other people)
 - 4. lack of social or emotional reciprocity
- B. Restricted repetitive and stereotyped patterns of behavior, interests and activities, as manifested by at least one of the following:
 - 1. encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
 - 2. apparently inflexible adherence to specific, nonfunctional routines or rituals
 - 3. stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements)
 - 4. persistent preoccupation with parts of objects
- C. The disturbance causes clinically significant impairment in social, occupational, or other important areas of functioning.
- D. There is no clinically significant general delay in language (e.g., single words used by age 2 years, communicative phrases used by age 3 years).
- E. There is no clinically significant delay 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.
- F. Criteria are not met for another specific Pervasive Developmental Disorder or Schizophrenia.

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Rett's Disorder

Rett's disorder was first reported by Dr. Andreas Rett in 1966. It primarily occurs in girls, whose very early development appears to be normal. Then, usually within the first year of life, head growth slows, interest in other people diminishes, and development in the areas of communication/language, thinking, and motor skills either stops (for very young children) or skills are lost. Generally, these children have profound intellectual disability and motor involvement, including distinctive repetitive hand-washing movements that are difficult to control. This rare disorder is caused by a mutation in the MECP2 gene on

the X chromosome (Autism Spectrum Disorders, n.d. Retrieved October 2007, from http://www.nimh.nih.gov/disorders/rett/detail_rett.htm). Rett's disorder is the only disorder in the pervasive developmental disorders category for which a specific cause has been identified.

Childhood Disintegrative Disorder (CDD)

Similar to Rett's disorder, childhood disintegrative disorder involves a period of normal development that is followed by regression, and is very rare. It occurs more often in boys than in girls. It was first described by Dr. Theodore Heller in 1908 (as cited in Volkmar, Koenig, & State, 2005). The regression starts between the ages of 2 and 10 years and is usually seen in language, social interest, cognition, toileting, and other abilities. Older children who have CDD appear similar to those with autism, but the early developmental history – with regression not occurring until the age of 2 years or later – is distinctive (Volkmar et al. 2005).

Pervasive Development Disorder not Otherwise Specified (PDD-NOS)

PDD-NOS is a diagnosis used for those who have marked impairment in social interaction or communication, and/or who display repetitive behaviors or intense interests, but do not meet the full criteria for one of the other pervasive developmental disorders or DSM-IV-TR diagnoses. For example, individuals with PDD-NOS may not meet the total of 6 of the possible 12 items under autistic disorder, may have symptoms that are milder, or may have first experienced symptoms after 3 years of age. As noted by Fillipek and colleagues (1999), PDD-NOS is a diagnosis by exclusion. There are many different ways a person might “not quite” meet the criteria for other ASD, so individuals with PDD-NOS comprise a rather diverse group.

Session 1, Lesson B. Autism Spectrum Disorders

Upon completion of this section, learners will:

1. identify the diagnoses that are included in the term “autism spectrum disorders” (ASD).
2. discuss the dimensions on which individuals with ASD differ.
3. describe the most recent prevalence estimate of ASD in the United States.
4. identify two proposed explanations for the increase in the prevalence of ASD.
5. discuss the male to female occurrence ratio in ASD.
6. describe the co-morbidity of ASD with intellectual disability and seizures.
7. identify a primary difference between what theorists thought caused autism in the 1950s-60s and today.
8. describe what is generally known about the inheritance of autism from twin studies.
9. identify three ways that the brains of children and adults with autism differ from those of typically developing peers.

The umbrella term “autism spectrum disorders” (ASD) is commonly used to describe a subset of the pervasive developmental disorders. The term *autism spectrum disorders*, first coined by Allen (1988), is not a diagnostic category but is widely used to refer to autistic disorder, Asperger’s disorder, and pervasive developmental disorder, not otherwise specified (PDD-NOS). The term “spectrum” appropriately implies the variability evident in individuals with autism spectrum disorders (ASD). Variability is expressed both in the severity of the symptoms seen from person to person, and in the range of cognitive abilities. Some individuals with an ASD have relatively mild symptoms of autism, while others are more severely impaired. Cognitive abilities range from severe intellectual disability to intelligence (IQ) in the genius range. These two dimensions – intelligence and severity of autism – do not necessarily co-vary in the same manner in individuals with ASD. For example, one person might have an IQ of 130 and have mild autism, while another person with an IQ of 130 may display more severe symptoms of autism.

The autism spectrum disorders are developmental disabilities, which are chronic conditions, typically lasting throughout a person’s lifetime. While the disability is life long, the specific characteristics and needs of individuals with autism spectrum disorders can change dramatically as they develop. In addition, many individuals benefit greatly from early identification and intervention services.

Prevalence, Gender, and Co-occurring Disorders

Prevalence

The autism spectrum disorders are among the most common of the developmental disabilities. The most recent prevalence figures in the United States come from studies conducted through the Centers for Disease Control and Prevention (CDC). In these studies, autism, Asperger’s disorder, and PDD-NOS were included as autism spectrum disorders. The CDC estimates that on average 1 in 110, or 0.9%, children in the United States have an Autism Spectrum Disorder (CDC, 2009).

Another recent estimate comes from reviews of the epidemiological literature by Fombonne (2005a, 2005b). He systematically considered epidemiological studies of autism spectrum disorder over about four decades. Using the studies published since 1987 to estimate “current” rates, Fombonne proposed an overall prevalence for ASD of 60/10,000 or .6%.

Is the prevalence of ASD increasing? This topic is hotly debated, and the answer is not entirely clear. Looking solely at the reported prevalence of autism over 4 decades, Fombonne (2005 a,b) cited a figure of 4.7/10,000 as the average prevalence rate of autism for studies published between 1966 and 1993. This contrasts with a much greater average figure of 12.7/10,000 from the studies published between 1994 and 2004. (Note that these figures are for autism, and do not include Asperger’s, PDD-NOS, or CDD.) Although it is clear that the reported proportion of children identified with

autism has increased, Sparacino, Noseworthy, Steiman, Reisinger and Fombonne (2010) suggest the increase in numbers may be due to “case definition, case identification, public awareness, social services and policies, and study design variables” (p.317). He also noted that most of the epidemiological data are not adequate to properly test for changes in the incidence of autism (Fombonne, 2005).

Gender, Race, and Social Class

From the earliest reports by Leo Kanner to the present, ASD have been observed to be more common in boys than in girls. In the CDC surveys, there were about four to five boys diagnosed with ASD for every girl (Centers for Disease Control and Prevention, 2009). This is similar to the ratio Fombonne (2005b) determined from his review in which he estimated an average male/female ratio of 4.3/1. Further, the male-to-female ratio is even higher for high-functioning autism and Asperger’s disorder, ranging from 6:1 to as high as 15:1 (Volkmar, Chawarska, & Klin, 2005)

Fombonne (2005b) also reviewed the few studies that have examined the association between race and autism and between immigrant status and autism. He concluded that the more methodologically sound studies have not found such associations. Similarly, he concluded that there does not seem to be an association between social class and autism.

Co-occurrence with Other Disorders

It is common for ASD to co-occur with other disorders; this is often referred to as comorbidity. The most common co-morbid conditions are intellectual disabilities (mental retardation) and seizure disorders. The CDC (2009) examined the cognitive characteristics, when available, of the children diagnosed with ASD in their surveys. Overall, between 29% and 51% had test scores indicating cognitive impairment (IQ ≤ 70). This proportion of individuals with ASD who have cognitive abilities in the average range is larger than the proportion reported in earlier studies. For example, a widely cited study by Gillberg (1984), conducted in Sweden, found only 23% of people with autism had IQs in the average or near-average range. Fombonne (2005b) cited three other studies published before 1986 that reported average cognitive scores in 16-30% of the individuals in their samples. It is likely that the shift – to a higher proportion of individuals with ASD having average or above average cognitive abilities – is associated with changes in the diagnostic criteria for autism and with the inclusion of Asperger’s disorder. Another factor could be more valid assessment of intellectual function in individuals with ASD (Johnson, Myers, & the Council on Children with Disabilities, 2007).

Children with ASD are at an increased risk for the onset of seizures prior to adulthood. Seizures develop in about one in four individuals with ASD (Tuchman & Rapin, 2002), with a peak onset prior to 2 years of age and again in adolescence. No one specific seizure type is characteristic for autism.

Some genetic syndromes have been reported to co-occur with autism. For example, fragile X syndrome, a genetic mutation involving a specific gene (FMR1), and tuberous sclerosis, a genetic disease that causes benign tumors to grow in the brain and other organs, are both associated with autism (see “Co-occurring Medical Conditions” section). A recent study of children with autism found a higher than expected prevalence of co-occurring mental health conditions including specific phobia, obsessive compulsive disorder, attention deficit hyperactivity disorder, and depression (Leyfer et al., 2006). In individuals with high functioning autism and Asperger’s disorder, anxiety and depression are common co-morbid conditions (Klin, McPartland, & Volkmar, 2005).

Etiology

Since Leo Kanner first identified the syndrome, our understanding of the causes of autism has grown considerably; nonetheless, there is still a lot that we do not know. Currently, there is considerable research underway to better understand the etiology (cause) and genetic components of autism, and more is being learned each year.

Early Theories

Many theorists in the 1950s and 1960s thought it likely that the causes of autism included a biological vulnerability and cold, non-responsive parents. Bruno Bettelheim was a well known and highly influential theorist in this period. His psychodynamic formulation of autism, as illustrated in his book, *The Empty Fortress: Infantile Autism and the Birth of the Self* (Bettelheim, 1967), was that parents, and especially mothers, were cold and rejecting of their children. Bettelheim’s belief that “refrigerator mothers” caused children to become autistic haunted parents for many years (Mesibov, Adams, & Klinger, 1997). In the 1960s and 1970s, however, evidence mounted to demonstrate that the differences seen in those with autism were biologically based and not caused by parenting.

Current Theories

It is now accepted that ASD are neurologically based developmental disorders (Bailey, Phillips, & Rutter, 1996). Most scientists believe that there are many different factors implicated in the etiology of ASD, although a specific cause can be identified in less than 10% of those who are diagnosed with ASD (Chakrabarti & Fombonne, 2005). Our knowledge about specific genetic patterns implicated in ASD is only in an emerging stage, but it is clear, as discussed below, that ASD is highly heritable (Bailey et al., 1996).

Co-occurring Medical Conditions

Autism has been found to be associated with many specific disorders or syndromes. The CDC estimates that about 10% of children who are diagnosed with an ASD also have an identifiable genetic, neurological or metabolic disorder, such as fragile X or Down syndrome (Kumar, 2009; Cohen, D et al., 2005). We know, for example, that autism occurs frequently in children who have fragile X syndrome with estimates

ranging from 25% to 67% (Hatton, Sideris, Skinner, Mankowski, Bailey, Roberts, et al., 2006), tuberous sclerosis (17%-64%), and untreated phenylketonuria (up to 5%) (Filipek, 2005). In children with Down syndrome, recent studies have suggested that 6%-7% also have ASD (Johnson, et al. 2007).

Genetics and ASD

Research has demonstrated that genetic factors play an important role in autism. Twin studies provided some of the first evidence for a genetic component to ASD. Among identical twins, if one child has an ASD, the other is also affected about 60%-96% of the time (Boyle, et al, 2005). On the other hand, the rate in fraternal (non-identical) twins when one child has autism is about 0-24% for the other twin. Estimates of recurrence risks, based on family studies of idiopathic (without a known cause) autism are approximately 5% - 6% when there is an older sibling with an ASD. This recurrence rate increases to approximately 8% when there are already 2 children with ASD in the family (Rutter, 2005).

It is unlikely that a single gene will be found that explains the inheritable patterns seen in children and adults with idiopathic autism. More likely, several susceptible (vulnerable) genes will be identified that contribute to the great variability in symptoms associated with the broad autism spectrum disorders (Rutter, 2005). Scientists at many research centers are studying the genetics of ASD, and knowledge about the various genes implicated in ASD is expanding.

Environmental Factors

There has been speculation about environmental factors that might play a role in the development of autism. For instance, an association between ASD and early childhood vaccines, particularly the measles, mumps, rubella (MMR) vaccine, was reported in the 1990s. In 2001, the Institute of Medicine reviewed population-based studies and concluded that there was no evidence of a causal association between the MMR vaccine and autism (Institute of Medicine, 2001)

Concerns have also been raised about the effects of environmental mercury exposure (including vaccines containing the mercury-based preservative thimerosal) on brain development in persons with developmental disabilities, including ASD. Mercury has long been known as a neurotoxin in its organic form that leads to motor impairment and visual and intellectual deficits. Using large data sets from the United States, Sweden, and Denmark, no consistent association has been found between thimerosal-containing vaccines and the prevalence of autism. In its report, the Institutes of Medicine (2004) concluded that the evidence favored rejecting the hypotheses that the measles, mumps, and rubella vaccine (MMR) and the mercury-based preservative in vaccines, thimerosal, play a significant causal role in the development of autism, although the report review committee did not rule out the possibility that vaccines might contribute to the disorder in a small number of individual cases. Nonetheless, a 2006 survey of parents of children with ASD revealed that 54% of parents believed that their child's ASD was caused by

immunizations. It was also found that 53% thought their child's ASD was caused by genetics (Harrington, Rosen, Garneco, & Patrick, 2006). There are research efforts underway to identify other possible environmental causes, including exposure to lead and other heavy metals (Johnson et al., 2007).

Structural Differences in the Brain

Regardless of etiology, scientists have discovered differences in the development of the brain in individuals with ASD, and in the brain's structures and functioning. It appears that in many young children with autism, there are very early differences in the way the brain grows and develops that have their origin in the prenatal period. For example, increased head size is a common feature in children with ASD, with 20-30% having macrocephaly or a head circumference that is more than 2 standard deviations above the mean (Nelson & Nelson, 2005). Findings are beginning to emerge that point to specific differences in areas of the brain in children with ASD when compared with typically developing children. For instance, specific areas of the brain (frontal, limbic, basal ganglia, and cerebellar regions) may differ (Sokol & Edwards-Brown, 2004). This is thought to account for some of the impairments in emotion, in social interaction, and in imitation. Differences in patterns of connections among cells in some parts of the brain may be linked to the difficulties in integrating information that are commonly seen in individuals with autism. Functional neuroimaging techniques have indicated the presence of abnormalities in face recognition and executive functioning in adults with high-functioning ASD (Brambilla, Hardan, diNemi et al., 2004). These and other findings will help us understand how differences in brain growth and development might account for some of the unusual features of autism as well as the variability that exists.

Additional information and a diagram of the effected brain structures implicated in autism can be found at www.nimh.nih.gov/publicat/autism.cfm, and www.nimh.nih.gov/health/publications/autism/research.shtml. Launch the video at this site, www.msnbc.msn.com/id/6827424, to learn more about how functional MRIs can assist in understanding the interactions among brain structures in individuals is ASD.

Session 1
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