Autism and Pervasive Developmental disorders

Carolyn R. Fallahi, Ph. D.
About Autism

• What do children with Autism look like?
  – Language
  – Unresponsive to people
  – Strange behaviors
  – Specific deficits

• The history of Autism
  – Changeling children
  – St. Francis of Assisi
  – Modern research: childhood psychosis
  – Kanner and Autism: infantile autism
  – New research into Autism
  – Asperger’s Disorder
Diagnostic Criteria

• The concepts of autism and PDD have changed in the various editions of the DSM.
  - DSM-II = childhood schizophrenia.
  - 1960’s and 1970’s (Rutter) = that autism is a separate disorder because it is different from childhood-onset schizophrenia (the current diagnosis).
  - DSM-III-R included 3 disorders in the PDD category; infantile autism, childhood-onset PDD (similar to Asperger’s Dx now), and atypical PDD.
Asperger’s

- DSM-IV: AD became a separate diagnosis and the age requirement for autistic disorder was reinstated – onset before 36 months; and 2 new disorders, Rett’s disorder and Childhood disintegrative disorder, were included.
Diagnostic Criteria

• There is controversy as to whether AD, CDD, and Rett’s DX should be separate and distinct disorders or combined into one broader autism spectrum disorder (ASD).
Autistic disorder

- **qualitative impairment in social interaction, as manifested by at least two of the following:**
  - 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
  - failure to develop peer relationships appropriate to developmental level
  - 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)
  - lack of social or emotional reciprocity

- **qualitative impairments in communication as manifested by at least one of the following:**
  - 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)
  - in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others
  - stereotyped and repetitive use of language or idiosyncratic language
  - lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level

- **restricted repetitive and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:**
  - encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus
  - apparently inflexible adherence to specific, nonfunctional routines or rituals
  - stereotyped and repetitive motor manners (e.g., hand or finger flapping or twisting, or complex whole-body movements)
  - persistent preoccupation with parts of objects

- 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.
- The disturbance is not better accounted for by Rett’s Disorder or Childhood Disintegrative Disorder.
Asperger’s Disorder

Qualitative impairment in social interaction, as manifested by at least two of the following:

– **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**
– **failure to develop peer relationships appropriate to developmental level**
– **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)**
– **lack of social or emotional reciprocity**

• Restricted repetitive and stereotyped patterns of behavior, interests and activities, as manifested by at least one of the following:
  – **encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity of focus**
  – **apparently inflexible adherence to specific, nonfunctional routines or rituals**
  – **stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements)**
  – **persistent preoccupation with parts of objects**

• The disturbance causes clinically significant impairment in social, occupational, or other important areas of functioning.

• 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).

• 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.

• Criteria are not met for another specific Pervasive Developmental Disorder or Schizophrenia.
Comparing Autism and AD

• Criteria similar between Autism and AD.
  – Deficits in social relationships
• Symptoms also include restricted, stereotyped behaviors as well as a need for sameness.
• Differences
  – Language development
  – No significant delay in cognitive development
• High functioning Autism
  – IQ>70
  – IQ<70
  – Spectrum disorder?
Evidence for Spectrum Model

• Norway study
• Miller & Ozonoff (2000) = neuropsychological tests to children with high-functioning autism and children with Asperger’s disorders.
• However….differences in outcomes that suggest the need for distinct disorders.
Does an individual child have a PDD?

• No biological markers
• The diagnosis of PDD is made solely on observations of a child’s behaviors.
• The Childhood autism Rating scale (CARS) used with preschool children.
• Differential diagnostic issues, e.g. MR vs. Autism.
• What symptoms are indicative of Autism? Aspberger’s disorder?
Impairments

- Attachment – Do they attach to parents?
- Specific deficits that interfere with social functioning.
  - Orienting their attention
  - Joint attention behaviors
  - Imitation
  - Difficulties processing visual stimulation for faces
  - Language communication problems
  - Deviant language
  - Restricted, repetitive, and stereotyped behaviors
  - Intelligence
  - Sensory perception
  - Self-injurious behavior
  - Aggression
  - Sleep problems
Asperger’s Disorder

- Similar diagnostic criteria except for language and cognitive development.
- Asperger’s disorder vs. high-functioning autism. Are they really different?
- Language problems that are evident in Asperger’s.
- Motor problems.
- Epidemiology.
Development during Childhood and adolescence

- Chronic disorder.
- The intensity of some symptoms, such as repetitive, stereotyped behaviors, may fluctuate while the intensity of other symptoms, such as language and social impairments are more constant.
- IQ is an important predictor of the type and severity of symptoms children with AD exhibit.
- Prognosis?
Affective Impairments as the foundation of ASD

- Hobson (1989) and Mundy & Sigman (1989) hypothesized that the fundamental deficit in autism is an affective deficit.
- Baron-Cohen (1988), Happe and Frith (1995), & others have hypothesized that the fundamental deficit in ASD is cognitive: Infants and young children with ASD do not comprehend that others have feelings and thoughts that differ from their own.
- Trepagnier (1996) and Smith and Bryson (1994) proposed theories that both focus on perceptual impairments as the fundamental problem in autism.
Etiology

• Neurological explanations
  – brain morphology (structure of the brain) & neurotransmitter functioning.

• Several NTs have been implicated:
  – Serotonin: linked to problems with sensory perception, motor function, and memory
  – Dopamine: restricted and stereotyped behaviors
  – Norepinephrine: attention, arousal, memory, anxiety, and movement
Genetics

• Twin studies.
  – 60% MZ twins concordant for autism
  – 5% DZ twins concordant
  – Heritability index: 90% - strong genetic influence
Vaccines

• Vaccines against measles, mumps, and rubella?
Rett’s Disorder

• All of the following:
  – apparently normal prenatal and perinatal development
  – apparently normal psychomotor development through the first 5 months after birth
  – normal head circumference at birth
• Onset of all of the following after the period of normal development:
  – deceleration of head growth between ages 5 and 48 months
  – loss of previously acquired purposeful hand skills between 5 and 30 months with the subsequent development of stereotyped hand movements (e.g., hand-wringing or hand washing)
  – loss of social engagement early in the course (although often social interaction develops later)
  – appearance of poorly coordinated gait or trunk movements
  – severely impaired expressive and receptive language development with severe psychomotor retardation
History

• 1960s Andreas Rett, an Austrian Psychiatrist.
• The diagnostic criteria for Rett’s disorder and childhood disintegrative disorder are similar, although there is more of an emphasis on problems with physical movement (esp hand movement) in Rett’s.
Rett’s disorder

- Normal development until approximately 5 months old.
- They look similar to low-functioning Autism.
- They have marked impairments of language and social interaction and engage in stereotyped behaviors.
- These behaviors are often limited to hand behaviors.
- IQ severe or profound range.
Rett’s Disorder

- Rett’s vs. other disorders
  - Breathing
  - Swallow air
  - Night laughter
  - violent screaming
Prevalence & Course

- 0.7 to 1 case/10,000
- Females only
- Genetic mutation?
- First sign of loss of functioning – after 5 months.
- The loss of previously developed skills occurs between 1 and 3 years of age for most children with Rett’s disorder. The loss is rapid in some children; slower in others.
- Language and social responsiveness lost.
Rett’s

• No purposeful functioning of hands.
• Stereotyped hand movements, such as hand wringing, twisting, clapping.
• Lost many gross motor skills, making walking and other large body movements difficult, breathing problems begin and increase in severity.
• Epilepsy
• Many adults cannot walk.
Etiology of Rett’s

• If MZ twin: 100% of the twin pairs concordant for Rett’s. No DZ twin pairs have been concordant. This suggests strong genetic influence or possibly exclusive genetic influence.

• Implicated genes? X chromosome?
Rett’s

- Gross motor problems
- Poor coordination and walk with awkward gait
- Deceleration in foot growth
- Often their legs and feet become locked in rigid positions
- Back deformities are common
- Confined to wheelchairs
Childhood Disintegrative Disorder

- Apparently normal development for at least the first 2 years after birth as manifested by the presence of age-appropriate verbal and nonverbal communication, social relationships, play, and adaptive behavior.
- Clinically significant loss of previously acquired skills (before age 10 years) in at least two of the following areas:
  - expressive or receptive language
  - social skills or adaptive behavior
  - bowel or bladder control
  - play
  - motor skills
- Abnormalities of functioning in at least two of the following areas:
  - qualitative impairment in social interaction (e.g., impairment in nonverbal behaviors, failure to develop peer relationships, lack of social or emotional reciprocity)
  - qualitative impairments in communication (e.g., delay or lack of spoken language, inability to initiate or sustain a conversation, stereotyped and repetitive use of language, lack of varied make-believe play)
  - restricted, repetitive, and stereotyped patterns of behavior, interest, and activities, including motor stereotypes and mannerisms
- The disturbance is not better accounted for by another specific Pervasive Developmental Disorder or by Schizophrenia
Childhood Disintegrative Disorder

• Theodore Heller in 1908 = dementia infantilis (dementia of the mind) to describe 6 children who developed normally for several years and then experienced an extreme loss of function.

• After the loss of abilities, the children look similar to low-functioning autistic disorder.

• Lowest functioning of all children on IQ – profound or severe range.

• Most are institutionalized.
Prevalence and course

- Only 126 cases of CDD had been identified by 2002, indicating how rare this is.
- Ratio of boys to girls is 4-5:1.
- Develop normally for 2 years.
- Little is known about the development.
Etiology

- Causes remain a mystery.
- CDD may represent a specific form of ASD, with a later onset and more severe course than the others.
- It also could be distinct. Not sure. Too small a number to really know.
Therapeutic Interventions

• Cannot cure autism or other PDDs.
• The goal = help patients to help their families cope effectively with having children with PDD.
• Early intervention.
• Ivar Lovaas and colleagues = therapy intervention.