

Autism: What it is, What it isn't

Autism Asperger Conference
Fall 2007

Nancy Minshew, MD
Director, Autism Center of Excellence
University of Pittsburgh

Prevalence of Autism Spectrum Disorders (ASD)

Approximately
1 in 166 children

Baird et al. 2000¹	Chakrabarti & Fombonne²	Brick Township, NJ³	Chakrabarti & Fombonne⁴
1/170	1/160	1/150	1/170

¹ Baird et al., 2000; ² Chakrabarti & Fombonne, 2001; ³ Bertrand et al., 2001;

⁴ Chakrabarti & Fombonne et al., 2005

Prevalence: World-Wide and PA

- Most recent study: 1% children born
- Verbal or high functioning
- If prevalence correct: >74,000 in PA
- Autism epidemiologists think increased diagnosis reflects:
 - improving recognition which is still poor
 - improving recognition of the entire spectrum, not just those in the middle of the spectrum
 - beginning recognition of older children & adults

Estimates of Expressive Language Level at Age 9

Studies of Chicago & North Carolina:

- 39.6 – 40.9% speak in complex sentences
- 28.9 – 35.3% speak in sentences, but not fluent
- 10.5-16.8% have words but not sentences
- 14.3-14.4 have no or few consistent words

Terminology

- Diagnostic and Statistical Manual of Mental Disorders-IV-TR (DSM-IV)
- International Classification of Diseases-10 (ICD-10)
- A neurological disorder
- Most services provided within mental health
- Double-edged sword
- Will address difficult behavior
- But does so without understanding its cause

Official Category in DSM-IV: Pervasive Developmental Disorder

- Autistic Disorder
- Asperger's Disorder
- Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS)
- Childhood Disintegrative Disorder (4-12 yrs onset) (CDD)
- Rett's Disorder

Informal Category Not in DSM-IV: Autism Spectrum Disorders

- Autistic Disorder
- Asperger's Disorder
- Pervasive Developmental Disorder Not Otherwise Specified
- Accurate distinctions between these outside a research setting unlikely. Need a functional definition in social, language, adaptive and problem behavior domains instead.

PDD-ASD

- CDD and Rett's are rare and degenerative, often associated with early death.
- Hence, you are unlikely to see them and they will not be discussed.
- The focus will be on ASD solely. The terms ASD and PDD can and are used interchangeably but when used typically refer to the first three diagnoses only and NOT CDD and Rett's.

Other Caveats About ASDs: Medical Causes

- In 90-95% of ASD cases, there is no known cause for them besides ASD
- 5-10% have an associated disorder that causes the ASD
- Most common above chance are fragile-x syndrome, tuberous sclerosis and chromosome 15q deletion syndrome

Misdiagnoses of Children With ASD

- Developmental Delay/ Mental Retardation (ID)
- Language delay
- ADHD
- Social emotional delay or disturbance
- Oppositional Defiant Disorder
- Conduct disorder
- Adjustment Disorder
- Bipolar disorder
- Obsessive compulsive disorder

Misdiagnoses of Adults With ASD

- Schizophrenia
- Personality disorders (schizoid, borderline)
- Bipolar disorder
- Obsessive compulsive disorder
- Learning disabilities
- Mental retardation
- Anything in DSM-IV, usually multiple

Bipolar Disorder: Not

- BPD diagnosis often made because of “mood swings” or outbursts; individuals says what they think unfiltered- angry at what they see as unfair or unjust- or react to bullying or to social intensity of the environment
- Their fundamental social impairment is not recognized and no one thinks of ASD; typically not able to work or get along in social settings; not drug responsive

Autism vs Schizophrenia (Teens)

■ Schizophrenia:

- self-talk (thinking out loud-not talking to voices)
- negative symptoms (worsening psychosis-not)
- disorganization of thinking (lack of conceptual framework or grounding- not circumstantial, circumlocutious)
- hallucinations- memories of past events
- preoccupation w/ right and wrong-religion (interpreted as hyperreligiosity)
- magical thinking (childlike) (delusions)

Autism vs Mental Retardation

- Often seen as two separate disorders
- Actually “mental retardation” is the more severe expression of the cognitive difference apparent in all people with autism
- Typical cognitive profile in autism: focus on details/ difficulty grasping concepts and meaning
- About 50-60% of people with autism have IQ scores below 70
- Almost all with autism have adaptive behavior scores in mental retardation range

Recognizing ASD

- Strange or odd: reflects social impairment
- Monotone voice: usually too loud
- Little to no facial expression
- Upset by change, rituals for doing things in set ways; scripts for saying things
- Obsessions- with collecting stuff or a topic; super memory for facts or attention to small details
- Clumsy, awkward

Other Distinguishing & Important Features

- No hallucinations
- Onset in first three years
- Socially emotionally very young: naïve, child-like
- Very poor perspective taking if any
- Poor face & emotion recognition
- Gullible
- Very few strategies for problem solving, not flexible

Asperger's Disorder vs Autism

- Absence of delayed & disordered language development now and before 5 years
- Fewer symptoms than for Autistic Disorder
- Often precocious language development
- Actual application of diagnosis: highly variable with poor distinction between HFA, AS, PDDNOS

Quick Diagnosis of ASD in LFA

- **Intermediate severity:** echolalic (uncontrollable & immediate repetition of words spoken by another), few scripted stereotyped sentences; socially isolated; self-stimulatory behavior; difficulty with change; sensory issues
- **Most severe:** no language, no comprehension, no prosody, no adaptive behavior- out of proportion to IQ; direct care staff can tell who has autism vs non-autistic MR- they are highly familiar with IQ expectations

Diagnostic criteria for 299.00

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. (*on the following 3 slides*)
- B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years:
- social interaction
 - language as used in social communication, or
 - symbolic or imaginative play

Diagnostic criteria for 299.00 Autistic Disorder (cont'd)

1. Qualitative impairment in social interaction
 - 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

Diagnostic criteria for 299.00 Autistic Disorder (cont'd)

2. Qualitative impairment in communication
 - 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

Diagnostic criteria for 299.00 Autistic Disorder (cont'd)

3. Restricted repetitive and stereotyped patterns of behavior, interests, and activities:
 - 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 mannerism (e.g., hand or finger flapping or twisting, or complex whole-body movements).
 - d) persistent preoccupation with parts of objects

Behavioral Expression of Social Deficit

- **Most severe:** no response to any social overture & no attempt to initiate social contact with others
- **Intermediate:** a few unvarying stereotyped ways of initiating contact, contact initiated solely for needs; not sustained; no understanding of social etiquette between people or in groups
- **Less severe:** interactions consist of monologues, extended scripts that sound original but aren't, or are dependent on others' questions; inadvertently offensive to others but also naive and gullible; can't chit chat

Language & Communication Deficits

- **Most severe:** global aphasia & aprosodia (loss or impaired ability to speak or understand language); mute
- **Intermediate:** echolalic with no comprehension → a few echoed sentences used functionally to indicate needs → a few stereotyped sentences used rotely with extremely limited comprehension; short scripts; talks but not to you
- **Less severe:** grammatically correct sentences but deficits in comprehension of idioms, metaphors, and stories; talk about obsessions; monologues; long scripts; unending questions; cannot chit-chat; conversation not reciprocal
- **Bottom line:** expression is greater than comprehension

Expression of Impairment in Play

- **Most severe:** complete lack of interest in toys
- **Intermediate:** interest in smell, taste, or texture of objects; preoccupation with the parts of toys (spins wheels, lines up; carries around): nonfunctional or atypical play
- **Less severe:** functional play sequences but play is stereotyped; may precisely imitate video or TV; preoccupied with game shows, letter-word games, computer, video games- advantage due to piecemeal processing & rule based

Restricted & Repetitive Behavior: Expression of Local-Global Cognitive Deficit

- **Most severe:** self-stimulatory behavior disproportionate to IQ, oblivious to change
- **Intermediate:** interest in elementary features but not whole, tantrums with change, rituals for doing things, controlling of what others do and say
- **Less severe:** tolerant of ordinary changes, narrow range of interests that are preoccupations or obsessions with a focus on details, no common sense or insight; poor concept formation, problem solving; inflexibility; rule and fact based

Social Emotional Immaturity: Disturbance in Affective Contact Not Included in DSM

- Capacity to experience, comprehend, and regulate emotions at a basic and cognitive level is severely impaired and unrecognized despite frequent abnormal imaging abnormalities of the amygdala, an emotion structure of the brain.
- Most verbal ASD adults are socially-emotionally 12-18 months to 4-5 years of age. Failure to recognize this in treatment worsens behavior.

Co-Morbidity Or Autism

- Hyperactivity: part and parcel of autism; EF
- “ODD”: resistance to change
- “OCD”: usually restricted and repetitive behavior; rarely it is co-morbid OCD
- Seizures: 30% in autism by third decade; gene
- Sleep disorders in 50%
- GI disorders 50%
- Eating disorders frequent
- Genetic Metabolic disorders rare but present

Motor Impairments

Part and Parcel of Autism

- Clinical motor tests commonly document impairments
- More rigorous testing rare except grooved pegboard for praxis and finger tapping
- Linked to poor sign language
- Postural control impaired- linked to multi-modal sensory integration impairment (vestibular-visual-position sense)
- No rigorous studies of gait done yet

Sensory Impairments: Part and Parcel of Autism

- Common also
- More common in children than adults
- Can be severely impairing
- Sound, light, tactile, taste sensitivity & aversions
- Also preoccupations
- Pain and temperature insensitivity
- Can be disabling and impact behavior
- Respect them