Agenda

- Prevalence
- Definition/Diagnostic criteria
- Neuroscience and Genetics
- Features/Red Flags
- Screening / Assessment
- Resources
QUIZ

Prevalence
Prevalence

- **Current Prevalence**
  - 1 per 88

- Male to female ~ 4:1

- The largest increases among Hispanic and African-American children.

CDC 2012
<table>
<thead>
<tr>
<th>Surveillance Year</th>
<th>Birth Year</th>
<th>Number of ADDM Sites Reporting</th>
<th>Prevalence per 1,000 Children (Range)</th>
<th>This is about 1 in X children...</th>
</tr>
</thead>
<tbody>
<tr>
<td>2000</td>
<td>1992</td>
<td>6</td>
<td>6.7 (4.5-9.9)</td>
<td>1 in 150</td>
</tr>
<tr>
<td>2002</td>
<td>1994</td>
<td>14</td>
<td>6.6 (3.3-10.6)</td>
<td>1 in 150</td>
</tr>
<tr>
<td>2004</td>
<td>1996</td>
<td>8</td>
<td>8.0 (4.6-9.8)</td>
<td>1 in 125</td>
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<tr>
<td>2006</td>
<td>1998</td>
<td>11</td>
<td>9.0 (4.2-12.1)</td>
<td>1 in 110</td>
</tr>
<tr>
<td>2008</td>
<td>2000</td>
<td>14</td>
<td>11.3 (4.8-21.2)</td>
<td>1 in 88</td>
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</tbody>
</table>
## Earliest Known ASD Diagnosis

### Median Age and Proportion by Diagnostic Subtype

ADDMN Network, 2008

(Combining data from 14 sites completing the 2008 surveillance year)

<table>
<thead>
<tr>
<th>Subtype of Earliest Diagnosis:</th>
<th>Autistic Disorder</th>
<th>ASD/PDD(^1)</th>
<th>Asperger Disorder</th>
</tr>
</thead>
<tbody>
<tr>
<td>Distribution of Subtypes:</td>
<td>44%</td>
<td>47%</td>
<td>9%</td>
</tr>
<tr>
<td>Median Age of Earliest Diagnosis:</td>
<td>48 Months</td>
<td>53 Months</td>
<td>75 Months</td>
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\(^1\) Autism Spectrum Disorders/Pervasive Developmental Disorder – Not Otherwise Specified

**Limitations:**

A. Diagnostic information obtained from evaluation records may not capture the exact age of each child's earliest diagnosis.
B. 20% of these children had a different subtype noted after the earliest diagnosis.
Definition/Diagnostic criteria
Definitions - OLD

Autism Spectrum Disorders

- Asperger’s Disorder
- Autism
- PDD NOS
  - Rett Syndrome
  - Childhood Disintegrative Disorder
Definitions - NEW

Autism Spectrum Disorder

- Asperger's Disorder
- Autism
- PDD NOS
- Rett Syndrome
- Childhood Disintegrative Disorder
Three areas of Interest

- Socialization
- Communication
- Stereotyped/Repetitive Behaviors
New DSM-V conceptualization
A. Persistent deficits in social communication and social interaction across multiple contexts

1. Deficits in social-emotional reciprocity
2. Deficits in nonverbal communicative behaviors used for social interaction
3. Deficits in developing, maintaining, and understanding relationships

Specify severity based on social communication impairments and restricted repetitive patterns of behavior
DSM-V Diagnostic Criteria

- B. Restricted, repetitive patterns of behavior, interests, or activities, as manifested by at least two of the following

  1. Stereotyped or repetitive motor movements, use of objects, or speech

  2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns or verbal nonverbal behavior

  3. Highly restricted, fixated interests that are abnormal in intensity or focus

  4. Hyper- or hyporeactivity to sensory input or unusual interests in sensory aspects of the environment

- Severity is based on social communication impairments and restricted, repetitive patterns of behavior
DSM-V Diagnostic Criteria

- C. Symptoms must be present in the early developmental period

- D. Symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning.

- E. These disturbances are not better explained by intellectual disability or global developmental delay.
Across multiple settings

- Home
- School/Day care
- With other adults

- Are there differences
- Are there consistencies

- In young children this can be limited
Why do we diagnose?

- Categorization of child’s presenting symptoms
- Provide an answer to parents
- **Informs interventions and educational services**
- Provides a universal language that specialists across disciplines can understand
- Allows for more specific communication with schools and other providers
- provides justification for increased and specialized services
Who Can Diagnose?

- Psychologist
- Developmental Pediatrician
- Neurologist
- Psychiatrist
When can we diagnose

- Age of first parent concern: 14-18 months
- Report symptoms to clinician: 18-26 months
- Lag time until diagnosis: usually after age 3
Earliest Known ASD Diagnosis
Median Age and Proportion by Diagnostic Subtype
ADDMN Network, 2008

(Combining data from 14 sites completing the 2008 surveillance year)

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Limitations:
A. Diagnostic information obtained from evaluation records may not capture the exact age of each child's earliest diagnosis.
B. 20% of these children had a different subtype noted after the earliest diagnosis.
BUT... We can make reliable diagnoses in Toddlers
Early predictors of ASD

- Successfully predicted over 90% of children with ASD at 14 months.
  - Poor or inconsistent response to name
  - Low frequency of babbling; low inventory of consonant sounds in communicative vocalizations
  - Limited response to joint attention
  - Limited initiation of joint attention
  - Receptive language delay on Mullen Scales
  - Stereotyped/repetitive interests and behaviors

Landa, Holman, Garrett-Mayer, 2007
Stability of diagnosis

- High stability of diagnosis in children over the age of 30 months of age
- Stability is slightly less in younger children
- Stability of diagnosis is affected by
  - Experience of the clinician
  - Severity of symptoms
- Instability is usually due to shift from autism to PDD NOS; occasional shift from PDD NOS to non PDD NOS
Difficulties in Diagnosis in young children

- Some DSM-IV criteria useful for 24 month olds, but not for 18 months and younger

- Still Researching how early symptoms map onto later symptoms

- Few tools for assessing autism under 18 mos of age (more research than clinical tools)

- Full range of symptoms may not be present <3 yrs of age
Difficulties in Diagnosis in young children

- Can be difficult to distinguish from:
  - Intellectual Disability
  - Language or global developmental delay
  - Trauma/attachment problems

- Rule out
  - Hearing loss
Difficulties in Diagnosis in young children

- Myths/misconceptions
- Parental/caregiver/provider denial
- Lack of attention to parental concerns
- Changing criteria
Concurrent Diagnoses with ASD

- Congenital hearing loss
- Developmental language disorder
- Genetic disorders: Down, Rett, fraX, Williams syndromes
- Tuberous sclerosis
- PKU
- Seizures
- GI disorders
- Macrocephaly: familial megalencephaly, hydrocephalus
- Tourette syndrome, Sydenham chorea
- Obsessive compulsive disorder
- Bipolar disorder
- ADHD
Neuroscience and Genetics
Neuroscience of Autism

- Brain based, neurological condition
- Compared to controls, ASD brains:
  - Early brain overgrowth
  - Differences in gray and white matter
  - Reduced synchronization between key brain region for different cognitive tasks
  - Processing connections within brain regions may be too strong

Williams, 2008
Both ASD groups:
• surplus of short-range connections within different brain region
• Relatively fewer connections linking far-flung areas
• The redundancy of pathways within brain regions is consistent with previous research a lack the normal "pruning" that normally takes place in infancy

Peters, et al., March 2012
Genetics of Autism

- 33 genes identified as having a role in Autism
  - Prior to that 11 genes identified
  - Changes in genes predisposes children to the development of Autism
  - Identified precise points where gene alterations take place
  - Identified genes also related to psychiatric disorders but with alterations in different places

Talkowski, 2012
New Baby Sibs Study

- Those with an Autistic child are at higher risk for having a second child with Autism
  - Girls are 9% more likely
  - Boys are 26% more likely
Insert cnn clip on sibling risk


Features/Red Flags - Social
Joint Attention

- Joint attention: shared attention/interest/enjoyment
  - Examples
    - looking into the direction another person is looking
    - following a pointing gesture
  - Examine both initiation of and response to

- Involves regulation and communication:
  - Trying to get the other person to do/stop doing something
  - Socializing engaging – commenting, labeling

Note: Mastery of joint attention reflects a child’s ability and motivation to share in mental states of others (theory of mind)
JA in children with ASD

- Children with ASD use gesturing to communicate for regulatory purposes

- In children with ASD in particular IJA for social purposes is impaired

- They typically lack use of gesturing for social purposes

- Children with ASD do not follow gaze or use the adult’s facial expressions to influence their own behavior (social referencing)
Social referencing

- 3-point eye gaze: coordinating attention between people and objects
  - Looks at mom, then object, then mom

- Shared affect: Ability to recognize the emotional state of others

- Examples:
  - when infants are faced with a new situation, they might look toward their mother for an indication of delight, anger or fear in her facial expression
  - Infants facial expression will mimic mom’s
Response to joint attention
Initiation of joint attention
Initiating joint attention with alternating gaze
Observable behaviors

- What might this look like?

- Can you think of any kids you know that fit this description?
Observable Behaviors

- When playing with caregiver does child look at what caregiver points to (car, pet, cup, ball)
- Doesn’t look at pictures adult point out when reading or at toys they point out
- Doesn’t make eye contact with caregiver
- Doesn’t interact with other children (siblings, neighbors, cousins) – look at them, talk to them, refer to them
- Doesn’t look to others to share emotions
Video Clip

- [http://autismspeaks.player.abacast.com/asdvideoglossary-0.1/player/autismspeaks](http://autismspeaks.player.abacast.com/asdvideoglossary-0.1/player/autismspeaks)
- Social #4
Features/Red Flags - Language
Communication deficits

- Delay in language
- Lack of desire to communicate
- Lack of nonverbal communication
- If speech is present: lack of communicative intent (may be scripted, stereotypic, echolalic speech)
Early communication deficits

- Abnormal reciprocal mother/infant vocalizations at 5-6 mo
- Babbling that does not increase in complexity
- Failure to respond to name
- Non-words with atypical phonation (e.g. squeals, growls, and yells)
- Delayed receptive and expressive language milestones
- Delayed and decreased use of gestures
  - Conventional gestures (e.g. showing, waving, pointing)
  - Symbolic gestures (e.g. nodding head, depicting actions)
  - Mostly primitive motoric gestures to communicate (e.g. contact gesture of leading or pulling another’s hand)
Early communication deficits

- Increased idiosyncratic or inappropriate means of communication
  - Self injurious behavior, aggression, tantrums

- Echolalia
  - Immediate (eg in response to questions)
  - Delayed (scripted verses reciting memorized dialogue)
  - Implies impaired comprehension

- Pronoun reversal (often “you” for “I” or “me”)

- Inconsistent use of words (“pop up” words)

- Labeling skills >> functional language

- Appearance of expressive>receptive skills, but not functional

- Hyperlexia-early, self-taught decoding of written language
Observable behaviors

- What might this look like?

- Can you think of any kids you know that fit this description?
Observable Behavior

- Caregiver may say child doesn’t talk, is hard to understand, or sounds strange
- Language may be more responsive than initiated or restricted for highly motivating items/events (requesting favorite toy, food)
- May have language but:
  - not use it socially with peers
  - it may be restricted to certain favorite topics or repetitive
  - Has trouble answering questions or having a conversation
Video Clip

http://autismspeaks.player.abacast.com/asdvideo glossary-0.1/player/autismspeaks

- Communication #3
Features/Red Flags – Repetitive Behaviors
Restricted, repetitive, stereotyped pattern of behavior, interests or activities

- Peculiar mannerisms
- Unusual attachment to objects
- Obsessions/compulsions
- Stereotypies (repetitive, nonfunctional, atypical behaviors such as handflapping)
Play skills

- Lack of understanding symbolic meaning of toys
  - Lack of/limited interest in toys
  - Focus on sensory features
  - Restricted range - lining, spinning, arranging, hoarding, carrying

- Solitary play
  - Parallel play and interactive physical play may develop

- Later can develop more symbolic play, but still limited
  - Shorter sequences
  - Limited creativity or variability
  - Repetitious, mechanical
  - Often precisely imitated from videos
  - Limited role playing, especially involving other children
Repetitive behavior and regulation

- Repetitive body movements are often self-soothing/calming

- Engagement in repetitive movements/engagement with toys in odd way often used in place of seeking social regulation with caregiver
  - If scared / excited experience it internally or with object rather than with person
Observable behaviors

- What might this look like?

- Can you think of any kids you know that fit this description?
Observable Behavior

- Keeps to him/herself
- Plays with same toys repetitively — stacks blocks over and over, rolls cars back and forth, spins parts, twists things
- Focuses on sensory aspects of items — rubs, licks, squeezes, staring at things
- Stereotyped movements (flapping hands, rocking etc,) maybe be infrequent and more pronounced when stressed or upset
Video Clip

- [http://autismspeaks.player.abacast.com/asdvideoglossary-0.1/player/autismspeaks](http://autismspeaks.player.abacast.com/asdvideoglossary-0.1/player/autismspeaks)
  - Restricted patterns of interest #4
  - Repetitive motor #2
  - Preoccupations #1
Things to look for in over time

- Lack of or limited eye contact
- Self-directed, hard to interact with
- Difficulty following directions
- Often non-responsive
- Difficulty responding to name
- Doesn’t interact or respond
Things to look for in over time

- Can answer questions but in a strange way
- Plays by himself
- Focused on favorite toy or activity - repetitive
- May be able to entertain himself with no interaction
- Or may be very disruptive and require a lot of supervision
Things to look for in over time

- May have intense tantrums OR be underresponsive
- May have sleep difficulties
- May be a picky or restrictive eater
Next steps
Screening | Assessment | Diagnosis
Early Detection Needed

- CDC, Healthy People 2020 objectives, and American Academy of Pediatrics all recommend children be screened by age 2.

- Early screening and diagnosis improves access to services during a child's most critical developmental period.
Healthy People 2020

- Increase the proportion of young children who are screened by 24 months
- Increase the proportion of children who get a first evaluation by 36 months
- Increase the proportion of children enrolled in special services by 48 months
Screening tools

- Modified Checklist for Autism in Toddlers (MCHAT)
  - [www.firstsigns.org/downloads/m-chat.PDF](http://www.firstsigns.org/downloads/m-chat.PDF)
Who can screen

- Pediatricians
  - Required at 18 and 24/30 months

- Early Intervention providers
  - Speech pathologist, special education, etc.

- Psychologist, Neurologist, Developmental Pediatrician
Comprehensive Assessment is Multidisciplinary

<table>
<thead>
<tr>
<th>Multiple areas of functioning</th>
<th>Multiple specialists</th>
</tr>
</thead>
<tbody>
<tr>
<td>- Speech and language</td>
<td>- Psychologist</td>
</tr>
<tr>
<td>- Social</td>
<td>- Speech therapist</td>
</tr>
<tr>
<td>- Adaptive</td>
<td>- OT/PT</td>
</tr>
<tr>
<td>- Cognitive</td>
<td>- Developmental Peds</td>
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<td>- Motor/sensory</td>
<td>- Educational specialists</td>
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<tr>
<td>- Medical information</td>
<td>- Social workers</td>
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<td>- Developmental history</td>
<td>- Neurologist</td>
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<tr>
<td></td>
<td>- Psychiatrist</td>
</tr>
<tr>
<td></td>
<td>- Parents</td>
</tr>
</tbody>
</table>
Functioning in other areas

- Important to understand other developmental skill levels
  - Cognitive delay will impact intervention
  - Level of language delay is will impact intervention
  - Adaptive functioning will inform intellectual disability diagnosis

- Important to understand strengths
  - e.g. is child more responsive to auditory or visual stimuli?
  - What are reinforcers and motivators for that child?
  - How/when do you see optimal performance?
What can you do?

- **Refer to:**
  - Infants and Toddlers (0-3)
    - Use the Maryland Physician’s Guide
    - Specify Autism concerns
    - Include copy of M-CHAT
  
  - ChildFind (3-5)
    - Parent has to call
    - Specify Autism concerns

  - Testing through Kennedy Krieger Institute, Center for Autism and Related Disorders

  - Audiological Assessment to rule out hearing loss
How to help parents with testing

- Talk to them before testing
  - Help them understand what testing is for
  - Listen to fears

- Follow up with them after testing
  - Listen to reactions
  - Discuss intervention options if known
  - Discuss Preschool/school options if age appropriate
References/Resources

- [www.firstsigns.org](http://www.firstsigns.org)
- [www.autismspeaks.org](http://www.autismspeaks.org)
  - [http://www.autismspeaks.org/family-services/tool-kits](http://www.autismspeaks.org/family-services/tool-kits)