9.85 Cognition in Infancy and Early Childhood

Lecture 14: Autism
Schedule

• **Monday, November 21st** -- mirror neuron debate; research drafts back
• **Wednesday, November 23rd** -- Happy Thanksgiving; no class
• **Monday, November 28th** -- statistical learning in infancy
• **Wednesday, November 30th** -- means-ends reasoning; tool use (Room 3015)
• **Monday, December 5th** -- causal reasoning 1: explanation, prediction, intervention and counterfactuals
• **Wednesday, December 7th** -- causal reasoning 2: assumptions, theories, and evidence
• **Monday, December 12th** -- conceptual change
• **Wednesday, December 14th** -- papers due; children and social policy; class party
Today’s schedule

- Mini-lab meetings (a la Liz)
- Language and thought debate
- Autism
Autism

- What do you know?
- What have you heard?
Autistic syndrome disorders

1. Diagnosis
2. Causes
3. Theory of mind
4. And?
Diagnosis

- 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 impairments in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body posture, 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 to other people)

- d) lack of social or emotional reciprocity (not actively participating in simple social play or games, preferring solitary activities, or involving others in activities only as tools or "mechanical" aids)
Diagnosis

(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
Diagnosis

• (3) restricted repetitive and stereotyped patterns of behavior, interests and activities, as manifested by at least two 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 mannerisms (e.g. hand or finger flapping or twisting, or complex whole body movements)

• d) persistent preoccupation with parts of objects
Diagnosis

• All children with autism have:
  – Communication disorders
  – Abnormalities of social interaction
  – Repetitive behaviors
Diagnosis

• Many children with autism have:
  – Abnormal sensory experiences
    • Sound of rain, telephone, vacuum cleaner, waves may be painful
    • Touch of clothes, light brush painful
    • But may be oblivious to traumatic injury (broken arm; head bash) or extreme cold and heat
  – Mental retardation (especially for language)
  – 1:4 develop seizures
  – Other disorders -- Fragile X; tuberous sclerosis; PKU; allergies
Diagnosis

• Some babies appear to have early abnormalities -- lack of normal attachment, dislike of touch …

• However, at least 20% of babies appear to experience “regression” -- normal development followed by loss of words, social contact around 18-months.
Diagnosis

- CHAT: Checklist for autism in toddlers
- Try it …
- High specificity but poor sensitivity
Diagnosis

• Autistic spectrum disorders -- Pervasive developmental disorders, not otherwise specified.
Asperger’s syndrome

A. Qualitative impairment in social interaction, as manifested by at least two of the following:
   • 1. marked impairments 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
Asperger’s syndrome

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 social interaction), and curiosity about the environment in childhood

F. Criteria are not met for another specific Pervasive Developmental Disorder or Schizophrenia
ASD

• Domain-specific deficit?
• Occurs more often in relatives of mathematicians and engineers
Domain-specific?

- Often contrasted with Williams syndrome
- 1:50,000
- IQ<70
- Very social
- Large and unusual vocabulary
• List 6 animals
• `weasel," ``newt," ``salamander," ``chihuahua" ``ibex" and ``yak."
Figure removed due to copyright restrictions.

[Contrast between visuo-spatial and language abilities in Williams Syndrome]
Domain-specific deficit

• However, contrast may be exaggerated.
• Many commonalities
• Language impairments even in Williams syndrome children.
• Retardation and sensory abnormalities common to both.
• Some children may have dual diagnoses.
Causes of autism

• “There is no known single cause for autism, but it is generally accepted that it is caused by abnormalities in brain structure or function.” (Autism Society of America)
Autism

• “Autism is not a disease, but rather a disorder in which there may be a number of different causal pathways. In the vast majority of cases, no specific underlying cause of autism can be identified. A variety of genetic, metabolic, infectious, and environmental factors may be important.” (Testimony from NIH to the Health and Human Services Committee, 2000)
Figures removed due to copyright restrictions. Please see:

Autism increasing?

• “Unfortunately, we do not have a definitive answer to the question of what the true incidence of autism is. Estimates vary widely, but recent studies suggest that as many as one in 500 persons may be affected by some form of autism. Recent reports suggest that the incidence of autism may be substantially increasing. It is not clear that the reported increases can be accounted for by improved or expanded diagnosis, or by the increasing availability of educational and other support services, although these are surely factors.”
Autism increasing?

• 1.5 million Americans
• Increasing at a rate of 10-17% year (MIT news office)
Autism and vaccinations?

• Five large epidemiological studies conducted in the United States, the United Kingdom, Denmark, and Sweden since 2001 consistently provided evidence that there is no association between thimerosal-containing vaccines and autism. Similarly, 14 large epidemiological studies consistently showed no association between the MMR vaccine and autism.

Autism and vaccinations?

Autism and genetics?

- Males four times more likely to be diagnosed with ASD than females
- Families with one autistic child have a 5% risk of having another child with autism.
- Concordance rate with identical twins = 60%
Autism and prenatal testosterone?

• High testosterone (sampled in the amniotic fluid during pregnancy) causes
  – reduced eye contact and
  – reduced vocabulary size at 18 months old
  – and reduced communication skills at 48 months old.

• Empathizing/systematizing theory?
Autism and Theory of Mind

- Remember -- this is a very partial view of autism.
- Most of the following studies refer to children with autistic syndrome disorders and normal or close to normal IQ’s …
- Otherwise, they are IQ-matched (and age-mismatched) or IQ and age-matched to children with other developmental disorders.
Autism and TOM

• TOM abilities preserved in autism
  – Recognize and attribute animacy and intention
  – Understand that desires cause emotions
  – Follow eye gaze -- know when someone is looking at them
  – Use see in spontaneous speech
  – Know what someone is looking at
  – Understand situations as cause of emotions
  – Can predict emotions based on desire
  – Can identify emotions from facial expressions.
    • Some difficulty with “social emotions” -- pride, shame -- longer delay, more scripted.
Autism and TOM

• Abilities impaired in autism
  – Poor joint attention
  – Poor gaze monitoring
  – No directing visual attention of others (point to get objects but not “protodeclaritively”)
  – Little or no pretend or symbolic play
Autism and TOM

• Evidence for mindblindness.
• Eye gaze
  – Eye-Direction detector (eye/object dyad)
    • Detects eyes or eye-like stimuli
    • Know whether eyes are directed towards it or something else.
    • Infers that the agent sees what it’s looking at.
  – Intentionality detector (eye/goal dyad)
    • Recognize things with eyes as potential agents, with goals.
  – Shared-attention Mechanism (you/me/object triad)
Autism and TOM

• Evidence for mindblindness.
• Eye gaze
  – Eye-Direction detector (eye/object dyad)
    • Mommy sees the keys.
  – Intentionality detector (eye/goal dyad)
    • Mommy might try to go to the keys.
  – Shared-attention Mechanism (you/me/object triad)
    • Mommy sees that I see the keys.
Autism and TOM

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[Group data illustrating focus on eyes versus mouth]
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[Scanning patterns in response to social visual versus verbal cues]
Autism and TOM

• Evidence for mindblindness.
• Failure to distinguish mental representations and reality
  – Jack is thinking about a dog.
  – Jill is holding a dog.
  – Who can pet the dog?
Autism and TOM

• Evidence for mindblindness?
• Failure to understand mental properties of the brain.
  – Keeping you alive, helping you eat, helping you move
  – Thinking, wishing, dreaming
Autism and TOM

• Evidence for mindblindness?
• Failure to understand where knowledge comes from.
• Alice looks in the box
  – Paul touches the top of the box.
  – Who knows what’s in the box?
Autism and TOM

• Evidence for mindblindness?
• Failure to understand deception.
  – Hide the penny
  – Autistic children may hide the penny in one hand, but leave the other open.
Autism and TOM

• Evidence for mindblindness?
• False beliefs
  – First order delays
  – Second order impairments and delays
Autism and TOM

• Pragmatics (Gricean implicatures)
  – Cooperative principle
    • Mom, there’s a party at Joe’s.
    • How about that research paper?
  – Maxim of quality
    • John has a Ph.D. but I don’t believe he has.
  – Maxim of quantity
    • Do you have 2 brothers?
    • Any more than 2?
  – Maxim of relevance
    • Is your mom there?
    • Yes
  – Maxim of manner
Other than mind-blindness?

- **Deficit of central coherence (Frith)**
  - Assemble puzzles without the picture
  - Remember strings of nonsense words almost as well as sentences
  - Can recite stories verbatim but may not be able to give you the “gist” of the story.

- **Deficit of executive function (Russell)**
  - Poor planning
  - Perseverative
More info …

• Autism and Developmental Disorders Colloquium Series
• "Identifying neurocognitive subtypes in autism"
• Professor Helen Tager-Flusberg, PhD
• 6:00 pm, Thursday, November 17, 2005
• MIT Building followed by a reception
It is generally accepted that autism is a complex spectrum disorder. Because of the inherent heterogeneity that characterizes autism spectrum disorder, the search for specific risk genes, replicable neuroimaging findings, or a unitary cognitive deficit have failed. I will present the case for taking a different approach to advance research on the genetics, neurobiology and cognitive aspects of autism arguing for the important of dissecting the phenotype of autism into meaningful subtypes on the basis of secondary symptoms. Three examples of subtyping will be presented: language, cognitive profiles, and executive functions.