

# SOCIAL COMMUNICATION: TYPICAL AND ATYPICAL DEVELOPMENT

H-126

6 November 2017

1

## What do we mean by communication?

- We can think of “communication” as consisting of gesture, speech, language and pragmatic language
  - **Gesture** is use of hand, arm, etc. movements to convey meaning (we can all think of gestures that do so)
  - **Speech** is the expressive production of sounds to produce words and phrases.
  - **Language** is the understanding and production of words, phrases and grammatical structures to effectively understand and convey messages
  - Includes **Pragmatics** (next slide)
    - Example of “deficit” in pragmatics: when my French classmate in University asked why I wanted her to “break a leg” when we were heading into an exam...

2

## What is pragmatics?

Knowledge of the appropriate use, purpose, or function of language for a variety of interactions within a variety of contexts and with a variety of people

- Focus is on the *function* of language not form
- Includes using language appropriately in social situations
- Includes understanding communicative *intent*
- Communicative intent can be conveyed non-verbally
  - through gesture
  - through facial expressions

3

---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---



---

## What do we mean by social communication?

- Components include:
  - social reciprocity
  - social interaction
  - social skills
- It is reasonable, then, to focus on the exchange of social information through both verbal and non-verbal channels.
- Because there are many courses on campus focused on language, in this lecture will focus on non-verbal channels

4

---



---



---



---



---



---



---



---

## Lecture Outline

- Gestural communication
- "Reading" speech and voice
- "Reading" faces and facial emotion
- Some disorders of social communication:
  - Developmental Prosopagnosia (inability to recognize familiar faces)
  - Disorders of attachment, in which there are deficits in social reciprocity and social communication generally (e.g., reactive attachment disorder)
  - Mental health disorders, such as
    - Psychopathy (adults)
    - Callous/unemotional traits (children)
    - Social anxiety disorders
    - Schizophrenia
    - bipolar
  - **Autism**

5

---



---



---



---



---



---



---



---

## Part I: Gestural Communication

- Long history of interest in link between gesture and language
- Months before infants use their first words, they communicate with caregivers via gestures
- Communicative gestures emerge as early as 8-10 months
  - Giving, showing, pointing
- These gestures considered *intentional communication* and may be used to direct and maintain caregiver's attention to particular object or referent.

6

---



---



---



---



---



---



---



---

## Gestural Communication, con't

- Importantly, these gestures predate and predict infants' oral language skills; indeed, infants' meaningful gestures have proven to be a more reliable predictor of later language than the oral language input they receive from caregivers (see Meredith Rowe, HGSE)
  - Further, infants' early pointing gestures predict
    - their first words
    - the size of their vocabulary
    - Onset of two-word combinations
  - Symbolic gesture use typically emerges between 13-16 months; not surprisingly, is strong correspondence between symbolic gestures and first words - e.g., former generally appear right before latter.
  - In adults, similar regions of the brain involved in use of symbolic gestures (e.g., pantomime threading a needle or holding finger to lips to indicate "quiet") and of spoken language
    - Superior and inferior temporal cortices (modality-specific to sound vs. gesture)
    - left-lateralized inferior frontal and posterior temporal cortex
  - *Thus, is some suggestion of common, modality-independent system for symbolic communication*

## Gestural Communication, con't

- Intriguingly, recent work by Kuhn, Willoughby, Wilbourn, Vernon-Feagans, & Blair (2014) report that individual differences in communicative gestures at 15 months predicted language development at age 2 and 3 years, which in turn predicted executive function skills at 4 years.
    - Thus, infants who use more gestures had better language and EF skills between the ages of 2 and 4 years
    - Note that it was *language* that mediated link between gesture use and EF skills

## Speech/Language Perception

- Hearing
  - Speech
  - language

---

---

---

---

---

---

---

---

---

---

---

---

---

---

---

---

---

---

---

---

---

---

---

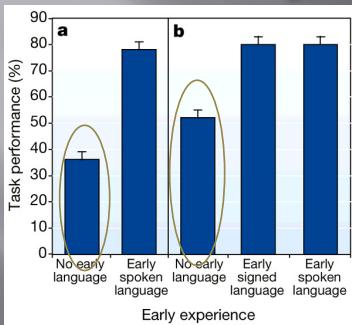
---

## Hearing Speech and Language

Nicholas, J. G., & Geers, A. E. (2007). *Journal of Speech, Language, and Hearing Research*

- Focused on language development among deaf children receiving cochlear implant
- Studied 3.5 and 4.5 year old children who had received implant by 3rd birthday
- Language scores increased with younger age at implant, even when controlling for duration of implant use.
- Children who received the implant at the youngest ages reached those of hearing age-mates by 4.5 years, but those children implanted after 24 months of age did not catch up with hearing peers.
- Conclusion:** Children who received a cochlear implant before a substantial delay in spoken language developed (i.e., between 12 and 16 months) were more likely to achieve age-appropriate spoken language. *Thus, there is a sensitive period for optimal recovery of hearing/language*

## Hearing, con't



Mayberry, R. L., Lock, E., & Kazmi, H. (2002). *Nature*

Language abilities of adults who learned English and/or American Sign Language (ASL) at different ages

**Study 1 (a) Groups:**

- Born deaf, no experience with language until learning ASL at school (age 9-15)
- Born hearing, English from infancy, learned ASL after becoming deaf at age 9-12

**Study 2 (b) Groups:**

- Born deaf, no experience with ASL until school (age 4-15)
- Born deaf, ASL in infancy
- Born hearing, English language in infancy, learned English at school age

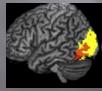
Conclusion: Language development compromised without early language exposure. Timing of early language influences later language learning capacity

## Hearing, con't

Some evidence suggests that even relatively brief auditory disruption during early language development may influence later language abilities

- Children who experienced multiple episodes of otitis media (ear infections) during their first years, resulting in periods of hearing loss, have shown subtle speech perception deficits into late childhood (e.g., Mody, Schwartz, Gravel, & Ruben, 1999)
- May be at risk for long-lasting central auditory impairments (Whitton & Polley, 2011)
- However effects are subtle and may depend on factors such as SES (e.g., Paradise et al., 1988), or the aspect of language that is measured (e.g., phonemic judgments vs expressive language)

A sensitive period for language in the visual cortex: Distinct patterns of plasticity in congenitally versus late blind adults



People blind from birth "hear" language in their visual cortex  
Thus, there is tremendous plasticity in sensory systems

Marina Bedny




---



---



---



---



---



---



---



---

## Speech and Language

- We don't recognize speech and we don't acquire language without hearing speech or language
- But as with vision, the brain likely comes pre-wired with the ability to benefit from experience
- With experience comes specialization

---



---



---



---



---



---



---



---

## Ontogeny of Speech/Language Perception

- Studies by deCasper and Fifer 30+ years ago established that infants' ability to recognize mother's voice due to exposure to that voice last weeks of pregnancy; thus, full term infants at birth
  - Recognize and prefer mother's voice vs. stranger's voice
  - Distinguish between mother reading familiar nursery rhyme (one heard prenatally) and unfamiliar
  - Do not recognize father's voice ☺

---



---



---



---



---



---



---



---

**A**

Adults

LH RH

- Dorsal pathway: Part of the AF/SLF connecting to Broca's area
- Dorsal pathway: Part of the AF/SLF connecting to precentral gyrus premotor cortex
- Ventral pathway: connecting the ventral portion of the inferior frontal gyrus to the temporal cortex via the extreme fiber capsule system

---

---

---

---

---

---

---

---

---

---

**A**

Adults

LH RH

**B**

Newborns

LH RH

Legend:

- Dorsal pathway: Part of the AF/SLF connecting Broca's area
- Dorsal pathway: Part of the AF/SLF connecting to precentral gyrus premotor cortex
- Ventral pathway connecting the ventral portion of the inferior frontal gyrus to the temporal cortex via the extreme fiber capsular system

---

---

---

---

---

---

---

# The timing and nature of experience with language affects perception of different languages

---

---

---

---

---

---

---

## Influence of Experience: Speech and Language Development

- ...but, if you give 9 month olds ~5 hours of exposure to non-native language (by live “tutor”), can recapture ability; if exposure occurs via audio or video tape, no effects \*

\*Kuhl, Tsao & Lie (2003) PNAS, 100, 9096-9101

---



---



---



---



---



---



---



---



---



---

## So, we have organization and reorganization




---



---



---



---



---



---



---



---

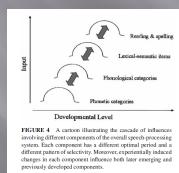


---



---

But a caveat: it may not be the case that there is **one** sensitive period for language; more likely is that there are multiple sensitive periods across development for different language domains



Werker & Tees (2005)

---



---



---



---



---



---



---



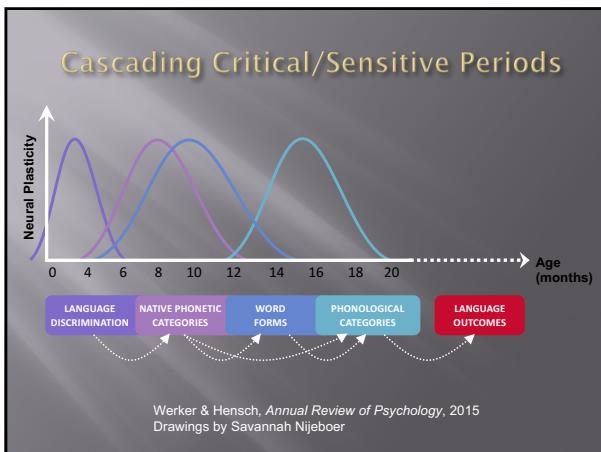
---



---



---




---

---

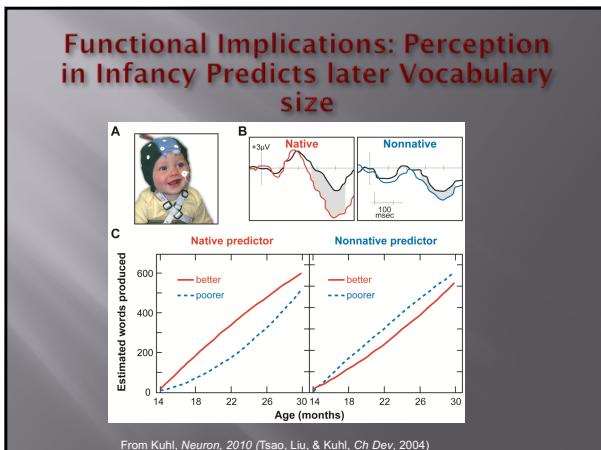
---

---

---

---

---




---

---

---

---

---

---

---




---

---

---

---

---

---

---

**Visual-only Language Discrimination**

- Filmed three bilingual French/English speakers



- Rendered into *silent* digital video clips
- Habituated infants to one language or the other
- Test with new vs original language




---



---



---



---



---



---



---



---



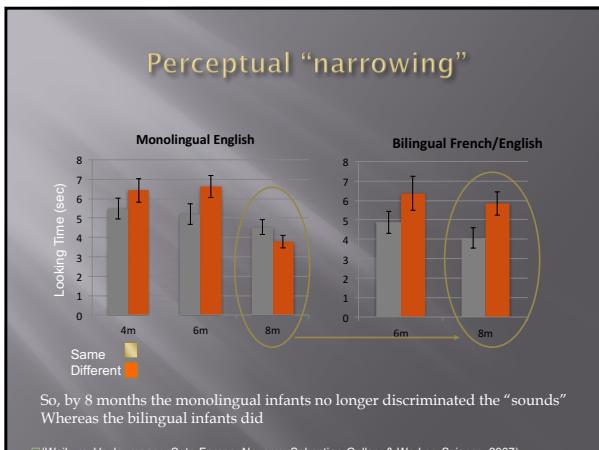
---



---



---




---



---



---



---



---



---



---



---



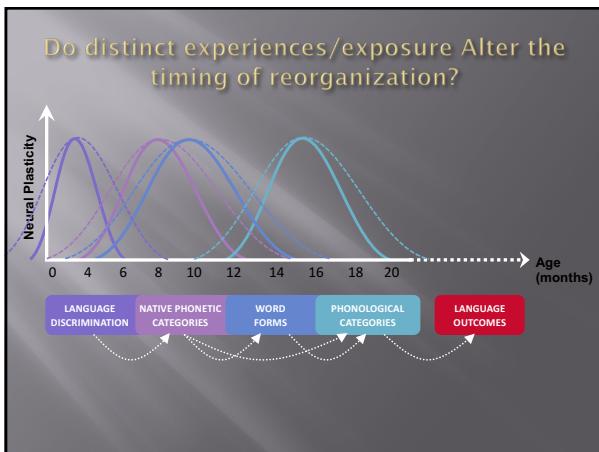
---



---



---




---



---



---



---



---



---



---



---



---



---



---

Language Group	Discrimination Score (S)
Spanish-Catalan (Monolingual)	~0.5
English (Monolingual)	~-0.5
Spanish-English (Bilingual)	~1.8
French-English (Bilingual)	~1.6

# Summary

## Part III: Reading Faces and Facial Emotion

## Are faces “Special?”

- Evidence in the adult that...
  - Behaviorally and neuropsychologically face and object processing are dissociable
  - Selective brain lesions impair face processing
  - In monkeys, there are specialized areas for processing faces and parts of faces
  - In the human adult, faces also appear to be processed in specialized areas, and
  - Face processing develops very early in life, suggesting, to some, that it is an experience-independent ability (what psychologists like to call “innate”)

31

---



---



---



---



---



---



---



---



---

## What is known about the behavioral development of face processing?

- Newborns
  - In some instances, prefer to look at faces vs. non-face objects
  - are able to discriminate faces from objects, but
  - there is no evidence that they see these as separate classes of stimuli; indeed, the preference for faces may be driven by the arrangement of facial features.
  - This preference is obtained using both faces (next slide) and dots (slide after next).

32

---



---



---



---



---



---



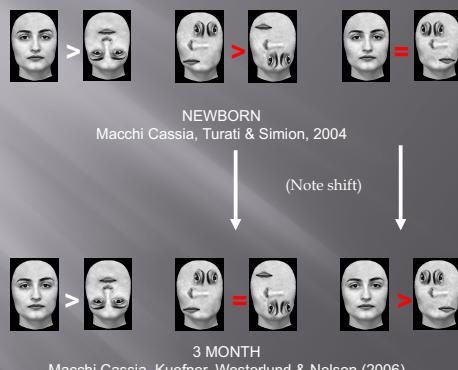
---



---



---



33

---



---



---



---



---



---



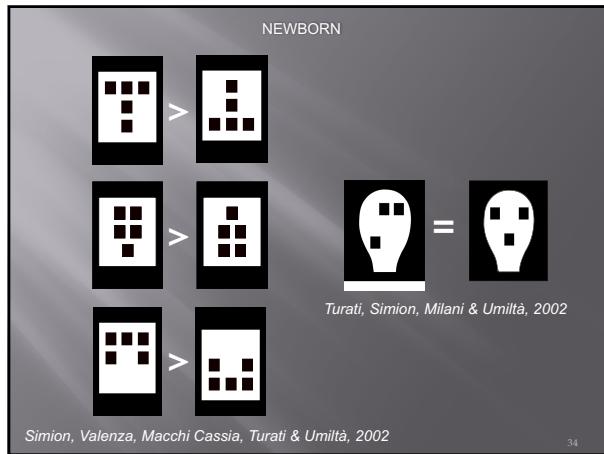
---



---



---



---

---

---

---

---

---

---

---

---

---

## What is known about the behavioral development of face processing (Con't)

- ~3-4 months infants begin to process faces as a distinct class of objects (e.g., they show the same inversion effect older children and adults do)
  - >4 months infants' face processing skills improve dramatically....although as we will discuss later, adult-levels of processing are probably not obtained until adolescence.

---

---

---

---

---

---

---

---

---

---

---

## Question 1: What do we know about Face v. Object Discrimination

- In infants, the N290 and P400 components of the event related potential (ERP) discriminates faces and objects
  - These components most prominent over posterior and temporal electrode locations (similar to adult N170) (see next slides)

---

---

---

---

---

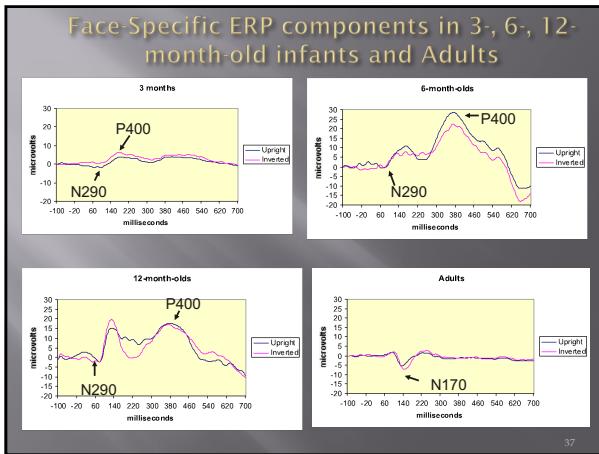
---

---

---

---

---



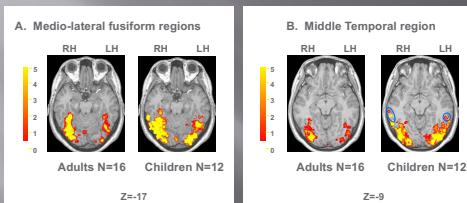
37

## What happens beyond infancy/toddlerhood?

- Even in childhood the neural systems that subserve face processing are more distributed than in the adult (see next slide)

38

## Face - matching Task



39

- Children, similar to adults, showed more right compared to left fusiform activation in response to faces, but activation in both hemispheres was more distributed across the temporal cortex in children compared to adults (Passarotti et al., 2003)

## How specific is face processing?

- Despite the improvements in face processing observed over the first 6 months, we must still ask how specialized and specific is this ability.
- One way this has been looked at is by examining the processing of other race faces and other species faces

40

---



---



---



---



---



---



---



---

## When does the other-species effect develop?

41

---



---



---



---



---



---

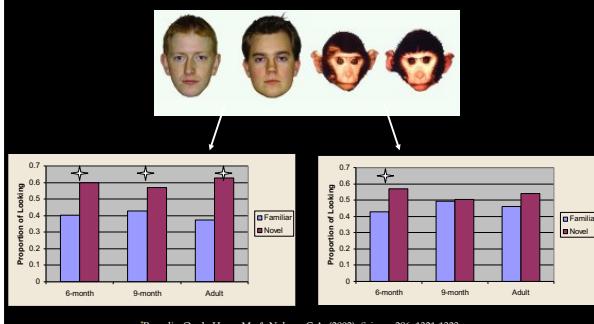


---



---

Pascalis, de Haan, & Nelson,  
2002



42

---



---



---



---



---



---



---



---

## Follow up Study

- Is it possible to keep open the perceptual window with additional experience?

43

---



---



---



---



---



---



---



---

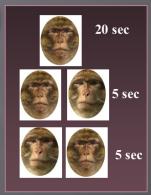
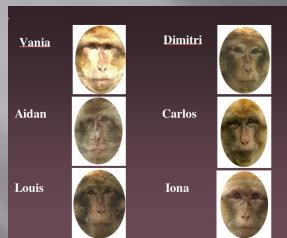


---



---

## Experiment 1: Training Infants



- Visit 1: 6-months
  - VPC
  - Sent home with monkey face book
- Visit 2: 9-months
  - Visual Preference (trained faces)
  - VPC (novel faces)

Pascalis, Scott, Kelly, et al., 2005

44

---



---



---



---



---



---



---



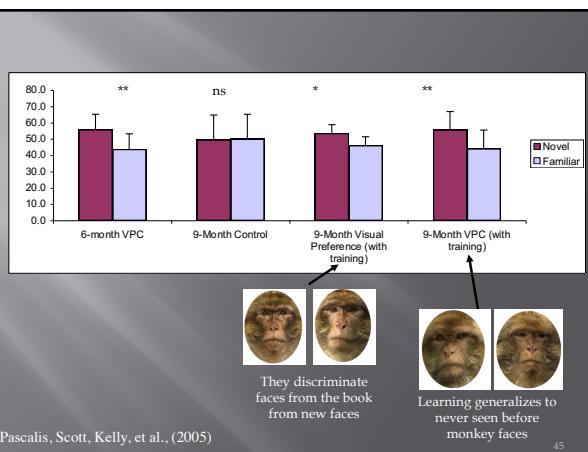
---



---



---



Pascalis, Scott, Kelly, et al., (2005)

45

---



---



---



---



---



---



---



---



---



---

## Pascalis et al. (unpublished)

- Performed adult study of monkey face discrimination
  - Group 1: adults with no experience viewing monkey faces received training equivalent to what infants received
  - Group 2: experts who held full time jobs working in monkey colony.

46

---



---



---



---



---



---



---



---

## Summary

- The “other species” perceptual window closes ~9 months, *unless* additional experience is provided.
- But, this experience must occur during a sensitive period; even adults who have extensive experience with monkey faces are not as good as infants if that experience was acquired as an adult

47

---



---



---



---



---



---



---



---

## How Important is Experience?

- Sugita (January, 2008: PNAS)
  - Reared monkeys from birth without access to faces (from 6-24 months)
  - When tested immediately after period of deprivation ended, all monkeys, regardless of deprivation, could discriminate monkey vs. monkey and human vs. human.
  - Then gave monkeys experience with either monkey or human faces; from that point on, could *only* discriminate faces from exposed category (lost the other)

48

---



---



---



---



---



---



---



---

## Arcaro et al. (2017)

- Extended work by Sugita; studied two groups of monkeys: reared from birth seeing faces and reared from birth with no face exposure
- Presented monkeys with images of faces, hands and objects while performing fMRI (monkeys >90 days old when scanning performed)
- Results: normal retinotopic organization for objects and hands but *not* faces
- Thus, similar to Sugita, find that exposure to faces is a requirement to develop face expertise

49

---



---



---



---



---



---



---



---



---

## Summary of Face Processing

- Collectively, it appears that the infant brain possesses a general neural substrate for all-purpose face processing, but face specialization is entirely (?) experience-dependent
- The perceptual window through which faces are viewed is broadly tuned early in life, and narrows with experience.
- Unknowns: a) nature of experience (what exactly must be observed to drive typical development?), b) timing (when must exposure occur?), c) duration ( how much exposure to faces required to drive system?).

50

---



---



---



---



---



---



---



---



---

## READING FACIAL EMOTION

51

---



---



---



---



---



---



---



---



---

Low hanging fruit:  
Is There a Differential Response  
to Negative vs. Positive Facial  
Emotion?

YES

- Evidence from looking behavior and EEG consistently finds that by 7 months infants "prefer" fear faces to happy faces (i.e., look longer at fear vs. happy; greater ERP response to fear vs. happy)

52

---

---

---

---

---

---

---

2009: At 7 months of age, infants exhibit adult-like attentional preference for fearful facial expressions

53

---

---

---

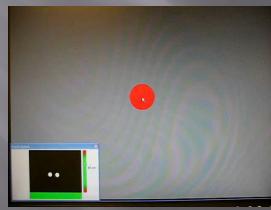
---

---

---

---

2009: At 7 months of age, infants also begin to exhibit adult-like attentional preference for fearful facial expressions



---

---

---

---

---

---

---

## Findings

- Infants take longer to disengage from fear face than happy face; i.e., attention to fear is prolonged
- This ability *not* present at 5 months but *is* present at 7 months
- Thus, is a perceptual bias to “prefer” fearful faces

55

---



---



---



---



---



---



---



---

## Example of reading emotion..



56

---



---



---



---



---



---



---



---

## Neural Bases

- The key components of the emotion-processing network and rudiments of the ability to recognize facial expressions emerge in early infancy
  - These components consist of:
    - Visual Cortex
    - Fusiform
    - Amygdala
    - Orbitofrontal Cortex

57

---



---



---



---



---



---



---



---

## What is Developing?

- Exposure to species-typical environment likely contributes to normal developmental trajectory;
- Conversely, deviations in this environment may derail typical development (e.g., maltreatment)
- Is a perceptual bias to attend most to fearful faces...and yet paradoxically
  - Despite longer looking and larger brain response, is no overt distress or discomfort and
  - recognition of fear does not reach adult levels of performance till adolescence.

58

---



---



---



---



---



---



---



---



---

## What is Developing? Con't

- Presumably basic neural architecture is constructed early, building both on experience with faces generally and later (when vision develops) facial expressions, and of course, on genetics
- System for perceiving and discriminating positive expressions likely in place very early, whereas negative expressions develops later....despite early bias towards fear

59

---



---



---



---



---



---



---



---



---

## General Conclusions About the Development of Face Recognition

- Evidence supports a rapid improvement in face recognition abilities through infancy, with more subtle changes occurring well into childhood.
- ERP and fMRI data also show changes in neural substrate across first decade of life.
- Presumably these changes reflect an experience-expectant system that is activity dependent ...and not any sort of "innate" module.
- There is probably a sensitive period (band width to be determined) associated with the development of facial expertise.

60

---



---



---



---



---



---



---



---



---

## Disorders in Social Communication

- Prosopagnosia
  - Inability to recognize familiar faces (see Oliver Sachs's "Man who mistook his wife for a hat")
  - Probably involves regions of inferior temporal and anterior occipital cortex, but evidence is a bit mixed
- Inability to read facial emotion, particularly fear
  - Patient S.M. Has genetic disorder leading to calcification of amygdala and she has gradually lost ability to recognize fear
- CASE STUDY: AUTISM

61

---



---



---



---



---



---



---



---



---

## DSM-V revision

### "Autism Spectrum Disorder"

- Includes autistic disorder (autism), Asperger's disorder, childhood disintegrative disorder and pervasive developmental disorder not otherwise specified

62

---



---



---



---



---



---



---



---



---

## DSM-V revision (con't)

### Must meet criteria A, B, C, and D:

- A. Persistent deficits in social communication and social interaction across contexts, not accounted for by general developmental delays, and manifest by **all 3** of the following:
1. Deficits in **social-emotional reciprocity**, ranging from abnormal social approach and failure of normal back and forth conversation through reduced sharing of interests, emotions, and affect and response to total lack of initiation of social interaction.
  2. Deficits in **nonverbal communicative behaviors** used for social interaction; ranging from poorly integrated verbal and nonverbal communication, through abnormalities in eye contact and body-language, or deficits in understanding and use of nonverbal communication, to total lack of facial expression or gestures.
  3. Deficits in **developing and maintaining relationships**, appropriate to developmental level (beyond those with caregivers); ranging from difficulties adjusting behavior to suit different social contexts through difficulties in sharing imaginative play and in making friends to an apparent absence of interest in people

63

---



---



---



---



---



---



---



---



---

## DSM-V revision (con't)

**Restricted, repetitive patterns of behavior, interests, or activities** as manifested by at least two of the following:

1. Stereotyped or repetitive speech, motor movements, or use of objects; (such as simple motor stereotypies, echolalia, repetitive use of objects, or idiosyncratic phrases).
2. Excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior, or excessive resistance to change; (such as motoric rituals, insistence on same route or food, repetitive questioning or extreme distress at small changes).
3. Highly restricted, fixated interests that are abnormal in intensity or focus; (such as strong attachment to or preoccupation with unusual objects, excessively circumscribed or perseverative interests).
4. Hyper-or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment; (such as apparent indifference to pain/heat/cold, adverse response to specific sounds or textures, excessive smelling or touching of objects, fascination with lights or spinning objects).

64

---



---



---



---



---



---



---



---

## DSM-V revision (con't)

- C. Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities)
- D. Symptoms together limit and impair everyday functioning.

65

---



---



---



---



---



---



---



---

## ASD

- Most often a lifelong disorder
- Rare that children “move off” the autism spectrum (2014 figures estimate 10% do so) although for many children, intervention (particularly early) greatly improves functioning
- ...and prognosis is improving

66

---



---



---



---



---



---



---



---

# The Autism Spectrum



---

---

---

---

---

---

---

## Other features

- Common co-morbidities
    - Behavioral difficulties (aggression, ADHD)
    - Psychiatric symptoms (anxiety, depression)
    - Epilepsy (about 1/3)
    - GI problems
    - Immune abnormalities
    - Sleep issues

---

---

---

---

---

---

---

# Cognitive Theories\*

- Theory of mind (TOM)
    - Deficits in the ability to infer the mental states of others
  - Central coherence (Uta Frith)
    - Deficits in information processing and integration
    - Excessive attention to detail, at the cost of the 'big picture'
  - Executive functioning deficits
  - 'Extreme male brain' (Simon Baron Cohen)

---

---

---

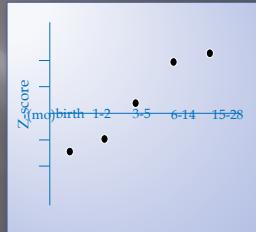
---

---

---

# Neurobiology

- Early studies in neuropathology and structural imaging, head circumference measurements
  - Most brain regions have been implicated (amygdala, frontal cortex, striatum, cerebellum)
  - Early dysregulation of brain growth
    - Rapid early growth (white and gray matter)
    - Plateaus at age 2-4
    - Likely affects long range connectivity,
  - 30% have macrocephaly



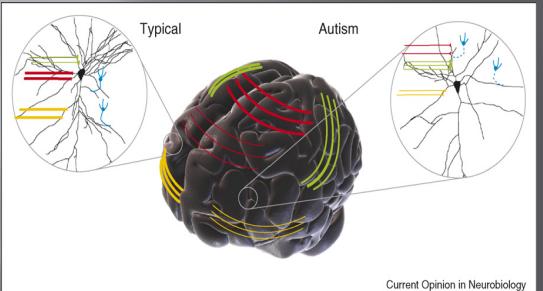
Courchesne et al JAMA 290: 337 (2003)

# Connectivity in ASD

- Field evolving based through functional and circuit based imaging
    - EEG (ERP, coherence), fMRI, tractography
  - Aberrant “connectivity”
    - local overconnectivity and regional underconnectivity
  - Likely problem of multiple circuits, not one structure



## Developmental disconnection in autism



Current Opinion in Neurobiology

## Demographics of ASD

- 70-85% of cases of autism associated with cognitive impairment
- Higher-functioning children being identified
- Why are the numbers going up?

73

---



---



---



---



---



---



---



---



---

## Current prevalence

- 1 per 68 for ASD (2014 figures)
- 4 boys:1 girl (but see subsequent discussion)
- Unclear race/ethnicity/cultural effects
- Regional differences (e.g., more common in New Jersey than in Alabama)
- Country differences

74

---



---



---



---



---



---



---



---



---

## Prevalence of Autism

Identified Prevalence of Autism Spectrum Disorder ADDM Network 2001–2012 Combining Data from All Sites				
Surveillance Year	Birth Year	Number of ADDM Sites Reporting	Prevalence per 1,000 Children Identified	This is about 1 in 68 children...
2000	1992	8	6.7 (4.1–9.3)	1 in 150
2002	1994	14	6.8 (3.1–10.6)	1 in 150
2004	1996	8	6.5 (4.4–8.6)	1 in 150
2006	1998	13	9.0 (6.2–12.8)	1 in 130
2008	2000	14	11.5 (8.4–15.7)	1 in 86
2010	2002	11	14.7 (10.0–20.0)	1 in 68
2012	2004	11	14.6 (10.7–19.0)	1 in 68

75

---



---



---



---



---



---



---



---



---

## Why the increase?

- Creation of an “autism” classification by Department of Education (DOE) in 1991
- ASD concept has broadened to include cases:
  - With less/no cognitive impairment
  - With milder symptoms
- Diagnostic substitution
- Earlier identification
- True increase in number of children with ASD?
  - Genetic vulnerability coupled with environmental “hit”?
  - But if that is true, what are the environmental contributions?

76

---



---



---



---



---



---



---



---



---



---

## What about gender differences?

- As a rule, males outnumber females 4:1. But
  - 6X as many males as females have normal intellectual ability, whereas this changes to 2X for those with moderate to severe intellectual impairment.
  - Thus generally, when girls have an ASD they are statistically more likely to be more impaired than boys
- Aggression, repetitive behaviors, restricted interests, reduced sociability, and hyperactivity generally occur more often in males
- Females generally have more anxiety, depression, and self-directed symptoms such as self-blame and low self-worth (of course, this is true outside of autism)

77

---



---



---



---



---



---



---



---



---



---

## Gender differences (con't)

- What about biological factors?
  - Relatives of females with autism are at increased risk for autism compared with relatives of males, suggesting that females with autism may carry a higher load of genetic risk factors, but conversely, certain biological aspects of “femaleness” protect against such variants (thus, the lower prevalence of autism in girls may be due to girls being “protected” from autism).
  - Genetic risk could be carried on X chromosome; thus, if one X has genetic variants, girls have a second X to counteract effect, whereas boys do not.
  - But, are likely other genetic protective factors (e.g., girls have fewer copy number variants than boys; i.e., fewer deletions or duplications of genes)

78

---



---



---



---



---



---



---



---



---



---

## Etiology of ASD

- About 15-20% due to known etiologic causes (e.g., Fragile X, tuberous sclerosis, etc.)
- Idiopathic ASD believed to have strong genetic origins (i.e., high heritability; e.g., 60-70%)

79

---



---



---



---



---



---



---



---



---

## Etiology of ASD

- Twin studies of autism

- Concordance for autism:
  - MZ: 36-91%; DZ: 0-30%
- Non-concordant co-twins (especially MZ) often have Broader Autism Phenotype (BAP)

(Example of BAP: "I see a family with 3 boys with ASD. I once saw them January 11, and recommended follow up in 3 months. They were scheduled for April 16. Mom emailed me to ask if they should be seen earlier, since the 3 month mark would have been April 11. Note that I have clinic once a week"

80

---



---



---



---



---



---



---



---



---

## Etiology of ASD

- Increased risk of diagnosis for siblings
  - (20%; vs ~1% in population)
- "Sub-clinical" features found in over 10% of sibs and 10-45% of parents (*Broader Autism Phenotype*)
- Affected siblings more similar to each other than to non-related child

81

---



---



---



---



---



---



---



---



---

## Etiology of ASD

→Multiple genes involved  
(at least count, ~800+ genes involved; see  
next slides)

→ Almost every chromosome has been implicated (but favorites include 15 and 16)

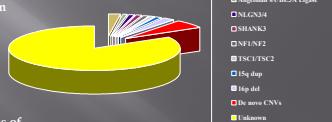
Known autism genes account for  
<20% of cases

- ▷ Deletions >20 genes (*in silico* de novo, generally)
    - 22q11, 16p11.2, others
  - ▷ Duplications
    - 22q11, 16p11.2, 15q11-13, others
  - ▷ Mental retardation syndromes with autistic symptoms
    - FMR1,UBE3A, TSC1/2, NF1/2, MeCP2
  - ▷ Rare, de novo, sporadic mutations cause mental retardation or autism
    - NLGN3/4, SHANK3
  - ▷ Impressions:
    - Highly heterogeneous
    - Each known gene typically causes  $\leq 1\%$
    - Majority unknown
    - Inherited factors largely unidentified for
    - Most mutations are partial loss of function
      - Hemizygous deletion
      - Heterozygous mutation

Category	Approximate Proportion
Known genes	Large slice (yellow)
Unknown	Medium slice (red)
De novo CNVs	Small slice (blue)
Other	Very small slice (green)

Reitveld et al., BEAS Ligue

  - Angelman syndrome
  - NLGN3/4
  - SHANK3
  - NF1/NF2
  - TSC1/TSC2
  - 16p dup
  - 16p del
  - De novo CNVs
  - Unknown



## Populations at High Risk for Developing an ASD

- Genetic syndrome / monogenic disorders, including
    - Tuberous Sclerosis Complex (risk of ASD= ~60%)
    - Fragile X (risk of ASD= 20-30%)
    - Rett Syndrome
    - Angelman Syndrome (risk of ASD= 50-81% )
    - Phelan-McDermid Syndrome (Shank3)
    - Down Syndrome (risk of ASD= ~5-15 % )
    - Copy number variants (e.g., 16P) (risk of ASD= ~20-30%)
    - Duchenne Muscular Dystrophy (risk of ASD=10-20%)

## Populations at High Risk for Developing an ASD

- Genetically-related syndromes:
  - Infants with an older sibling (risk of ASD=~20%)
- Non-Genetic pathways
  - Low birth weight (~20%???)
  - Ventriculomegaly
  - Infants raised in institutions (5-10%)

*Thus, there are many ways to "catch" autism*

85

## So....

- Could this possibly be the same disorder?
- Behaviorally, yes, they all these populations meet the same criteria
- ...but still, mechanistically, they must be different disorder
- Analogy: two people can have elevated blood pressure or body temperature, but the underlying causes are likely very different, as are the treatments (so, we do not have a "body temperature syndrome" or a "blood pressure syndrome")
- Thus, define the disorder behaviorally, which may be part of the problem

86

## Age of recognition

- Usually 15-18 mos
  - 1/3 within 1<sup>st</sup> birthday
  - Another 1/2 by 2<sup>nd</sup> birthday
- Epidemiological: well within first 3 years

87

## Age of diagnosis

- Around 4-5 years old
- Reliable diagnoses can be given by 18-24 months of age by “expert” clinician
- Earlier intervention = better results
  - Brain plasticity

88

---

---

---

---

---

---

---

---

## Earliest abnormalities

- Parent report
  - Language delay
  - Decreased social responsiveness
  - Nonspecific difficulties
    - Medical
    - Feeding
    - Sleeping
    - Motor delays

89

---

---

---

---

---

---

---

---

## Educational strategies

- Educational placements
  - Specialized settings
  - Inclusion settings
- Individual and group therapy sessions
  - Speech therapy
  - Physical therapy
  - Occupational therapy
  - Social Skills Group

90

---

---

---

---

---

---

---

---

## Educational strategies

- Individualized behavioral interventions
  - E.g., Applied Behavioral Analysis (ABA), Floortime, social groups
  - Conducted in AND out of school
    - Home
    - Specialized center
    - Community
  - General recommendation is total of 40 hrs/week

91

---

---

---

---

---

---

---

## Educational strategies

- Psychopharmacological interventions
  - Do not treat core symptoms
  - May ameliorate co-morbid symptoms/disorders
    - Seizures
    - Anxiety
    - Depression
    - OCD
    - Attention difficulties
- Note that for genetic causes of autism (e.g., TSC, Rett, etc) there are now a variety of clinical trials in progress

92

---

---

---

---

---

---

---

## Diagnostic Assessment

- Diagnostic Information
  - Interview with parents
    - developmental history
    - pervasiveness of behaviors
  - Assessment of Child
  - Observation in relevant environments

93

---

---

---

---

---

---

---

## Measures used for diagnosis

### Autism measures:

- ADI-R
- ADOS
- AOSI
- Checklists:**
- Social Responsiveness Scale
- CBCL or BASC

### Other measures:

- Vineland-II (Adaptive skills)
- Cognitive Measures
- Language and communication measures

94

---



---



---



---



---



---



---



---



---



---

## ASD specific measures

### Interviews:

- Autism Diagnostic Interview - Revised (ADI-R)
- Diagnostic Interview for Social and Communication Disorders (DISCO)
- Development, Diagnostic and Dimensional Interview (3di)

95

---



---



---



---



---



---



---



---



---



---

## ASD specific measures

### Observations:

- Autism Diagnostic Observation Schedule (ADOS)
- Autism Observation Scale for Infants (AOSI)

Note: clinical diagnosis (by pediatrician, psychologist) often does NOT depend on these measures

96

---



---



---



---



---



---



---



---



---



---

## Conclusion

- Although autism is many things, a primary feature is a problem in social communication
- Although everyone with autism has a problem in social communication, not everyone with a problem in social communication has autism; hence DSM 5 has new disorder *Social (Pragmatic) Communication Disorder*
- **Question:** under what circumstances should we consider autism a disorder vs. a difference?

97

---

---

---

---

---

---

---

---

THE END

98

---

---

---

---

---

---

---

---