Autism

(From the Diagnostic and Statistical Manual of Mental Disorders, 5th Edition)

1. A total of six (or more) items from (A), (B), and (C), with at least two from (A), and one each from (B) and (C)

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

1. impaired eye gaze or orientation or social responsiveness
2. failure to develop, maintain, or utilize social skills such as appropriate eye contact, sharing social interests, or attending to social overtures
3. marked delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate for this through alternative modes of communication such as gesture or mime)

(B) Qualitative impairments in communication, as manifested by at least one of the following:

1. delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate for this through alternative modes of communication such as gesture or mime)
2. abnormalities of speech initiation, rate, or rhythm
3. stereotyped and repetitive use of language or idiosyncratic language
4. abnormal echoic speech in response to questions or other discomfort

(C) Restricted or stereotyped patterns of behavior, as manifested by at least two of the following:

1. preoccupations with parts of objects
2. apparently inflexible adherence to specific, nonfunctional routines or rituals
3. repetitive and restricted use of language or idiosyncratic language
4. vivid, idiosyncratic or circumscribed interests
5. markedly restricted and repetitive motor mannerisms (e.g., hand or finger flapping or twisting, or complex whole-body movements)
6. repetitive and restricted patterns of behavior or use of objects, such as unprovoked head-banging, rocking, or head- or body-thumping

II. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years:

1. Language
2. Social interaction
3. Play

III. The disturbance is not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder.

Autism

The practical knowledge necessary to use and interpret language appropriately in social and real-world contexts.

Why are social contexts important?

How do you answer the telephone?

Why are real-world contexts important?

e.g., deictic terms, “here” vs. “there”

Pragmatics

Nonverbal gesture

Proto-declarative gestures (used to share interest in an object, or direct attention to an event) virtually absent (proto-imperatives normal)

Speech acts impaired

Declarative statements, showing off, acknowledging a listener, requesting information virtually absent

Conversational discourse

Deficits in conversational ability

Prosody

Deficits in pragmatic uses of prosody (e.g., non-grammatical stress, pauses)

All share an emphasis on social use of language!

Formulaic speech

What is a formula? A prefabricated sequence that is stored and retrieved whole from memory (“How are you?”: “You’re welcome”).

Formulas are important for social use of language; may also reflect a shortcut to avoid grammatical processing.

Types of formulaic speech in autism:

- Idiosyncratic sound-meaning associations (“I want to go blue” = go outside)
- Excessively literal language (“No, it’s raining water”)
- Trouble with pronouns and other deictic terms (you, me, here, this, etc.)
- Immediate/ delayed echolalia (lexically, prosodically, syntactically faithful)
- Abnormalities in intonation, voice quality (pitch), prosody

On average, compared to other children, children with autism have:

Higher rates of formulaic speech
Lower rates of spontaneous (non-formulaic) utterances
Syntax in Autism
- Spontaneous speech shows reduced syntactic complexity (relative to typically-developing children and developmentally delayed children)
- Closed class items omitted (e.g., the)
- Lower rates of novel, non-imitative utterances – higher rates of reliance on formulaic speech
- Impaired on tests of immediate sentence repetition
  - Impairments not found for all children!

Phonology in Autism
- Many studies report normal performance for individual speech sounds (for both expressive, receptive language)
- Some impairments found for combinations of sounds into syllables and words
- Deficits in repetition of auditorily presented nonsense words (barrazon)
- Non-word reading is less clearly impaired

Morphology in Autism
- Omission of inflectional morphemes in spontaneous speech in children with ASD
  - Produce play for playing, played, plays
- Irregular inflections relatively spared
- For elicited forms –
  - High rates of omissions and incorrect inflections in language impaired children with ASD (compared to children with ASD who had apparently normal language)
  - Wash or washing for washes, catch or catching for caught
- A different study found normal accuracy for children with ASD (relative to typically-developing children), but faster-than-normal response times for regular past tenses (walked, plugged, digged), but normal response times for irregular past tenses (dog, splim → splam).

Lexical Abilities in Autism
- Relatively intact word learning
  - Forming a sound-meaning association
  - Normal receptive vocabulary processing
    - Word-picture correspondence (“Is this a …?”)
    - Word-picture matching (choose the correct picture)
    - Picture selection (show me all the …)
    - Word definition (“What is a …?”)
- Spared single word production
  - Picture naming
  - Synonym/antonym generation
  - Reading single words out loud
- Occasional impairments on verbal fluency tasks
  - Rapid automatic naming (name pictures as rapidly as possible)
  - Name as many words as you can that start with the letter “f”
  - Name as many animals as you can

Conceptual Knowledge in Autism
- Seems to be largely spared
  - For individual word meanings
  - For conceptual organization of meanings
    - Children with ASD show normal pattern of prototypicality ratings for members of categories at both basic and superordinate levels
    - Semantic priming appears normal (based on one study)
  - Processing of words related to mental or emotional states seem to be impaired

Theories of Cognition in Autism
- Language and non-language domains

Category Labels
- Super-ordinate
  - mammal, animal
- Basic-Level
  - dog
- Sub-ordinate
  - beagle, terrier, rottweiler, alsation
Theory of Mind in autism

Theory of Mind hypothesis: Autism involves difficulty interpreting behavior as causally linked to mental states (Baron-Cohen, Leslie and Frith, 1985)

- Speech that does not entail viewing people as mental beings not necessarily impaired
- Speech that does entail viewing people as mental beings or requires shared attention (e.g., deictics) is impaired

Hypothesized to underlie both social and pragmatic impairments

Procedural Deficit Hypothesis

- Brain structures subserving procedural memory are dysfunctional
  - Frontal/basal-ganglia circuits
  - Frontal/cerebellar circuits
  - Predicts variability across individuals
  - Predicts range of grammatical abilities
  - Hypo (like Parkinson’s)
  - Hyper (like Huntington’s)
  - Compensation in declarative memory is expected
    - Strong lexical abilities
    - Over-reliance on formulaic speech

Procedural memory in autism

- Procedural Memory Functions
  - Impaired procedural learning
    - Rotary pursuit
  - Motor impairments
    - Impaired pantomime and imitation
    - Impaired complex skill learning (dancing)
    - Hypokinetic movements (bradykinesia)
    - Hyperkinetic movements (chorea)

Declarative memory in autism

- Declarative memory functions
  - Spared semantic memory
    - Normal representation and organization of word meanings
    - Strong ‘role’ memory
    - Normal paired associate learning
    - Impaired episodic memory

Complex Information Processing Deficit

- Across domains, complex functions impaired, simple functions spared
  - Language
    - Spared lexical processing (simple)
    - Impaired grammar/pragmatics (complex)
  - Non-language (e.g.)
    - Spared simple motor (finger tapping)
    - Impaired complex motor (grooved pegboard)
    - (complexity not well defined…)
**Weak Central Coherence**
- Superior performance in autism
- Strong performance on block design
- Inferior central coherence
- Superior local coherence

**Strong performance on Superior performance in**

**Savant Syndrome**
- Exceptional skills in context of impaired cognition
  - Prodigious – skills are exceptional compared to anyone
  - Kim Peek; Daniel Tammet
  - Talented – remarkable for age and cognitive level
- Close association between autism and savantism
  - Nearly every savant is on autistic spectrum
  - Nearly 10% of individuals with autism have savant abilities

**Summary of Theories**
- Impaired theory of mind
  - Explains pragmatic deficits
  - Says nothing about grammar or lexicon or motor or memory
- Procedural Deficit Hypothesis
  - Variation in grammar; spared lexicon
  - Links language to motor, memory performance
  - Says nothing about pragmatics
- Complex information processing deficit
  - Spared at simple functions (words, tapping)
  - Impaired at complex functions (grammar, pragmatics, motor)
  - Complexity not well defined
- Weak central coherence
  - Focus on smaller language units (words)
  - At expense of larger units (sentences, discourse)
  - Says nothing about motor, memory performance

**Domains of Savant Talent**
- All savants have exceptional memory
- All have obsessive-compulsive tendencies
- Some savants have talents in multiple areas

**Theories of savant talent**
- Excessive practice
  - Attentional deficits lead to excessive focus
  - BUT, some talents appear spontaneously...
- Exceptional rote memorization
  - Drawing, ability to play music not just memorization
  - Creation of a rich knowledge base through implicit learning
  - Vague on details of how implicit learning happens
  - Compensation
  - Right hemisphere compensates for damaged left hemisphere
  - Procedural memory compensates for dysfunctional declarative memory
  - Declarative memory compensates for dysfunctional procedural memory
- Stronger than normal episodic memory
- Extensive reliance on hippocampus for savant talent
- Possibly due to damage to the amygdala
- Weak central coherence
  - Focus on details leads to excessive practice, creation of rich knowledge base

**Studies of the Brain in Autism**
- Music
  - Perfect pitch
  - Hear a piece once, play it perfectly
- Art
  - Drawing / Sculpture
- Mechanical ability
- Memory
  - Personally experienced events; dates
  - Perfect memory for books (Kim Peek)
- Calendar calculation
- Arithmetic
- Language
  - Hyperlexia
  - Poetry
  - Language learning
  - Christopher Tietze
- Perfect pitch

**Summary of Studies**
- Autism and savantism
- Impaired cognition
- Strong central coherence
- Weak central coherence
- Exceptional rote memorization
- Excessive practice

**Exceptional skills in context of impaired cognition**
- Kim Peek; Daniel Tammet
- Talented – remarkable for age and cognitive level
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Brain in autism

- Few consistent findings
  - Abnormalities of Broca's area
    - Reduced volume
    - Reduced functional activation
    - Abnormal asymmetry (increased rightward)
  - Abnormalities of temporal lobe areas
    - Increased volume
    - Increased functional activation
    - Abnormal asymmetry
  - Reduced numbers of Purkinje cells in cerebellum
- Findings from other structures not consistent
  - Hippocampus
  - Basal-ganglia

A Structural imaging study of language in Autism (De Fossé et al., 2004)

Assessed language abilities of boys with autism, using CELF (Clinical Evaluation of Language Fundamentals) and non-word repetition

Examined cortical (grey matter) asymmetry in Broca's area (pars opercularis + pars triangularis) and planum temporale

Abnormal asymmetry in Broca's (R > L)
- Combination of decreased LH volume / increased RH volume
- Abnormal planum temporale asymmetry (L > R)
- Combination of increased LH volume / decreased RH volume

fMRI Study of Language (Just et al. 2004)

Visual sentence comprehension:

The comprehension task was to read a brief or passive sentence and respond to a probe. (Highlights on a separate line) identifying either the agent or the recipient of the action by pressing the left or right hand response button that was made inside the correct response, such as:

The cook thanked the father.
What does increased / decreased activation mean?

Results

- In ASD participants:
  - Normal accuracy
  - Faster than normal response times
  - Greater activation (than controls) in posterior superior temporal sulcus relative to controls
  - Reduced activation (compared to controls) in left inferior frontal gyrus; supplementary motor area
- What does increased / decreased activation mean?
Brain Growth in Autism (Redcay and Courchesne, 2005)

Summary

- Autism is complex, difficult to characterize
- A complete theory of autism needs to explain both strengths and weaknesses in the disorder
- Brain basis of the disorder is not well understood