Language acquisition in pathology (1)  

Autism (and SLI).


1. Autism is characterised by:

a. Profound withdrawal from human relationships; obsessive preoccupation with objects and set routines (cf. 'Rainman'), leading to repetitive and stereotyped patterns of behaviour, interests and activities
b. Impairments in communication, social interaction and emotional response: autists are ‘systemising’ rather than ‘empathising’ (Baron-Cohen et al, 2005).

2. These characteristics are standardly attributed to a lack of understanding of the mental states of others, due to a defective theory of mind (See Baron-Cohen, 1995; Tager-Flusberg, 2004; Smith et al, 2003, references therein; and (4) below). Most autistic children have subnormal intelligence (see Fay, 1993:191) but some are very 'high functioning' (Asperger’s Syndrome) e.g. Temple Grandin (1986); Donna Williams (1992, 1994, 1996).

3. Language deficits constitute a fundamental feature of autism. Only half the autistic population has any “functional language”, where this is defined as the ability “to produce at least some two-word phrases” (Tager-Flusberg, 2004:34). Linguistic characterization:

a. Mutism – either from lack of language or the ‘decision’ not to use it; Segmental phonology of autists is less impaired than their intonation and prosody, and may fall within normal limits. It is often better in echolalia than in spontaneous speech. Echolalia – (uncomprehending?) repetition of verbal utterances (occurs in 75% of autists). “What are you doing?” elicits the response: “What are you doing?” or just “Doing”. Pronoun reversal (or repetition). “You want candy” intending to convey “I want candy”. Common with 25% of typically developing children, but persists with many autists. Use of neologisms – “all the rest are grinkafups”. Communicative incompetence. Even when the grammar is as close to normal as can be tested, the use of language – especially utterance interpretation- is abnormal. (See (4)).

b. When language does develop, it emerges late. In extreme cases (see Windsor et al, 1994) some aspects of language may emerge after prolonged mutism; but this development is probably not a function of the language faculty. ‘Ann’ used “articles, plural markers, prepositions and other functional grammatical forms. But her ... spontaneous responses were generally one- and two-word functional utterances, for example 'Sue hit', 'Sue haircut'” (Windsor et al. 1994:100), and her maximal responses were 5 to 6 morphemes. Typical examples of her speech include: "the black is pants", "the cut putting bandaid", "The is makeup", "Opposite new old".
4. The communicative incompetence of high-functioning autists is attributed to a problem with Theory of Mind (ToM) and 'Metarepresentation' ("Mindblindness", see Baron-Cohen, 1995). If pragmatics is "a variety of mind-reading" (Wilson, 2003:109) then impairment of ToM will lead predictably to pragmatic problems. Typically developing children acquire ToM by around four years; autists may never do so.

a. Interpretation of picture sequences (mechanical, behavioural, mentalistic\(^1\)) (Baron-Cohen et al, 1986): 3 groups (Down syndrome, TD, Autistic – mental age 3+).
   - Mechanical: autists better than the other two groups
   - Behavioural: all three groups comparable
   - Mentalistic: autists worse than other two groups [group data].

b. The imputation of (false) belief and the recognition of intention. Sally-Anne, Smarties. First-order and second-order ToM tasks. If children reply correctly to the question "Where will Sally look for the sweet?" they display first-order mind-reading ability; if they reply correctly to "Where does Anne think Sally will look for the sweet?" they display second-order ability. Someone capable of attributing first- but not second-order mental states would be able to identify a speaker's informative intention (to make mutually manifest the information X), but not their communicative intention (to inform the hearer of her intention to make X mutually manifest). Hence the impenetrability to autists of small talk, where only the second-order communicative intention is relevant. (see Happé, 1993).

c. This account generalizes to all instances of 'Interpretive use' (Sperber & Wilson, 1995:224ff.. Simile, metaphor, irony, jokes, metalinguistic negation, rhetorical questions, use/mention, lying ... (see Smith & Tsimpli, 1995, for the case of C).

5. Heterogeneity in language ability in autism: (Tager-Flusberg, 2004). In a population of 89 children (80 boys, 9 girls; ages 4-14; IQ range 25 – 141) with ‘functional’ language:
   - Articulation skills: within normal range (cf. 3a)
   - Grammar and semantics (CELF\(^2\)): the most impaired
   - Vocabulary and Non-word repetition: below average but better than CELF

6. The difference between psychological theories (including linguistic ones) and genetic aetiologies. The latter is in general too gross to distinguish among the former.

i. What is the genetic basis for autism? (It is unlikely to be simple as impairments in social cognition and language acquisition dissociate genetically).
ii. Is it the same as, or overlapping with, that for SLI or other conditions?
iii. Can the genetic determinants of the linguistic problems of autists (and those with other conditions) be distinguished from those affecting non-linguistic ones?

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\(^1\) The task is to put in order a series of pictures telling a story: e.g. a balloon blowing away and getting burst (‘mechanical’), a child going to a shop and buying some sweets (‘behavioural’), a child expressing surprised dismay at the surreptitious theft of his sweet (‘mentalistic’).

\(^2\) Clinical Evaluation of Language Fundamentals
7. The genetic basis: Autism has high heritability allied with heterogeneous aetiology. “More than 2 and as many as 100 genetic variants may contribute to susceptibility to developing autism” Veenstra-VanDerWeele & Cook (2003:116). Familial studies may be misleading: most autistic people do not have autistic parents because most autistic people don’t reproduce.

a. MZ twin of an autist has a 60% chance of being autistic
DZ twin of an autist has a 4.5% chance of being autistic (≈ non-twin sib)
Member of general population has a 0.2% chance of being autistic

b. Chromosomal abnormalities have been located on 2q37, 7q, 22q13 and (mainly) maternally inherited 15q11-13. 7q and 2q are more relevant for the linguistic deficits found in autistic spectrum disorders (i.e. relevant dissociations are identifiable).

8. Parallels with SLI (and dyslexia). Recall that SLI is heterogeneous and is broadly classified into at least three subtypes (apart from “gSLI”): phonological disorder, expressive-language disorder, and mixed expressive- and receptive-language disorder.

a. The pattern in (5) mirrors the pattern found in SLI, especially as regards NWR and morphosyntax. “…language-impaired children [with autism] performed in strikingly similar ways to children with SLI” (Tager-Flusberg, 2004:41).

b. Bartlett et al (2002:51) report that the 13q21 region has also been implicated in autism. Importantly, the linkage analysis for 13q21 increased when population selection was made on the basis of language delay. See also Bartlett et al (2004) whose title says it all. (They again found significant linkage on 13q21).

c. Linkage analysis of families with SLI showed a locus for poor non-word repetition skill on chromosome 16q, and for expressive language disorder on chromosome 19q (Bishop, 2003:159). NB there are likely to be hundreds of genes close to a linkage site. (See The SLI Consortium, 2002.)

d. Note that the KE family’s SLI is atypical, and no other family has FOXP2 affected (see Newbury et al, 2002). However, SLI is associated with regions “in the vicinity of FOXP2” (O’Brien et al, 2003).

e. Dyslexia shows significant comorbidity with SLI. (SLI Consortium, 2002.)

9. Conclusions:

a. Both autism and SLI (and dyslexia) are phenotypically heterogeneous and have a substantial genetic component to their aetiology.

b. There is some slight overlap in the linguistic manifestation of the two conditions, but considerably greater overlap in the genetic loci associated with each. It is not yet clear whether this overlap includes much/any identity of individual gene disruption.
c. The contribution of genes to the (in)correct development of the language faculty is essentially unimpeachable, whether any such contribution is exclusive to language is still unclear.

Additional references:


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